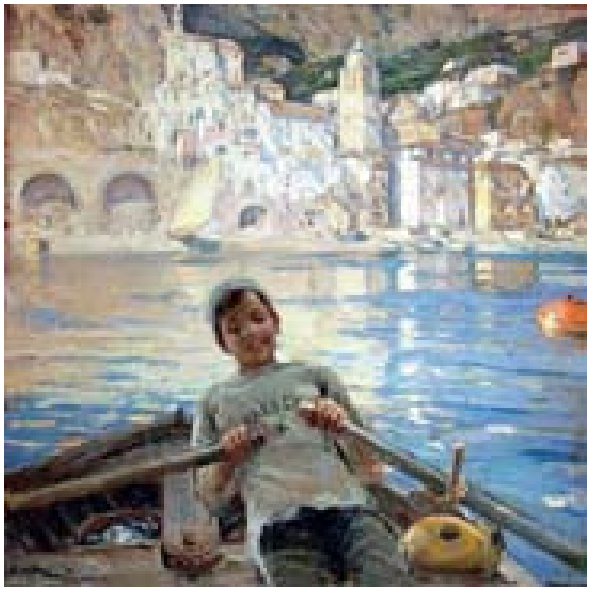
Malattie rare in età pediatrica:
Ricerca clinica, farmaci orfani e reti europee di riferimento



A cura della
Società Italiana di Ricerca Pediatrica

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Prefazione

Generoso Andria
Presidente SIRP

A distanza di un anno dal Forum sul futuro della ricerca clinica (non solo pediatrica)*, la SIRP, in collaborazione con la FIARPED e la SIP, propone una nuova riflessione, focalizzata sulla ricerca nel campo delle malattie rare in età evolutiva.

Le malattie rare rappresentano un settore della medicina non solo di crescente interesse per il sistema sanitario, ma anche molto fertile per l'attività scientifica, sia con lo studio delle loro basi fisiopatologiche, sia con lo sviluppo di approcci terapeutici innovativi attraverso sperimentazioni cliniche di farmaci orfani.

In coerenza col servizio che la SIRP intende svolgere attraverso il suo Osservatorio della ricerca pediatrica italiana, è stato organizzato A Napoli, nei giorni 29 e 30 giugno 2017, un Forum che rappresenta l'avvio di un monitoraggio dell'attività scientifica dei centri italiani e della loro partecipazione a *trial* clinici nel settore delle malattie rare in età evolutiva. È stato quindi impostato un catalogo sullo stato della ricerca pediatrica per le patologie rare, coinvolgendo numerose società scientifiche e gruppi di studio per le aree specialistiche di competenza, che monitorerà le attività rivolte alle patologie rare. Sarà un *work in progress*, aperto alla collaborazione della comunità scientifica, non soltanto pediatrica, per correzioni, integrazioni, commenti.

E quasi pleonastico sottolineare che anche per questo gruppo di malattie la qualità dell'assistenza dipende dalla qualità della ricerca scientifica e clinica. Da pochi mesi si è arrivati alla istituzione di reti europee di riferimento (ERN) per oltre 20 gruppi di malattie rare e ci è sembrato utile fare anche il punto sulla partecipazione alle ERN dei centri pediatrici italiani, di fatto cooptati come membri, tra l'altro, per l'eccellenza della loro produttività scientifica. L'auspicio è che analoghe reti per le varie specialità si possono costituire, o rafforzare se già esistenti, anche in Italia.

Un commento finale sul quadro scelto come logo del Forum e come copertina di questo Libro bianco. Il ragazzo che, da solo, rema con foga e con un sorriso verso la spiaggia inondata dal sole, illustra efficacemente quanto, come medici, auguriamo ai nostri piccoli pazienti: che assumano sempre maggiore responsabilità, in prima persona insieme alle famiglie, nella gestione della loro malattia, per raggiungere l'approdo di migliori condizioni di salute e qualità di vita.

Napoli, 29 giugno 2017

**** Il futuro della ricerca clinica (pediatrica), Giannini Editore, Napoli (2016)***

Guida alla lettura dei database

1. Lavori registrati in PubMed e pubblicati a partire dal 1 gennaio 2014 ad oggi per Malattie Rare dell'area specialistica con ruolo *leader* dell'Unità operativa (primo autore e/o autore corrispondente)

Istruzioni per la compilazione della tabella

1: indicare il nome della malattia o del gruppo di malattie rare oggetto del lavoro
2: indicare i nomi di tutti gli autori **(in grassetto il nome del primo autore e/o dell'autore corrispondente)**
3: indicare il titolo completo del lavoro
4: indicare il nome della rivista, il volume e le pagine
5: indicare l'anno di pubblicazione
6: indicare l'Impact Factor della rivista (scrivere **0,** se non assegnato)
7: scrivere eventuali note

1	2	3	4	5	6	7
Nome MR	**Autori**	**Titolo del lavoro**	**Rivista, volume, pagine**	**Anno**	**I. F.**	**Note**

N.B. nella versione online è possibile effettuare delle ricerche con parole chiave (autori, riviste, società scientifiche etc.)

2. *Trial* clinici per farmaci orfani per Malattie Rare dell'area specialistica con partecipazione dell'Unità operativa

Istruzioni per la compilazione della tabella

1: indicare il nome della malattia o del gruppo di malattie rare, oggetto del *trial*
2: indicare il nome dell'industria o dello *sponsor* indipendente
3: indicare con **I** se Internazionale**,** con **N** se Nazionale (scrivere **X** nella casella corrispondente)
4: indicare il ruolo dell'U.O. nel trial con **C** se Coordinatore, con **P** se Partecipante (scrivere **X** nella casella corrispondente)
5: indicare la del *trial*
6: indicare il nome del farmaco orfano sperimentale
7: indicare con **C** se il *trial* è ancora in corso, con **T** se il *trial* è terminato dopo il 1 gennaio del 2014, scrivendo in parentesi l'anno della fine (scrivere **X** nella casella corrispondente)
8: scrivere eventuali note

1	**2**	**3**		**4**		**5**	**6**	**7**		**8**
Nome MR	***Sponsor***	***Partners***		**Ruolo**		**Denominazione del *trial***	**Farmaco**	**Durata**		**Note**
		I	**N**	**C**	**P**			**C**	**T (anno)**	

N.B. nella versione online è possibile effettuare delle ricerche con parole chiave

TAVOLA ROTONDA

La ricerca clinica italiana per le malattie rare in età pediatrica

(in collaborazione con la rivista* Prospettive in Pediatria*)

Moderatore: Generoso Andria

Partecipanti: Renza Barbon Galluppi
Bruno Dallapiccola
Lucia Monaco
Ugo Capolino Perlingieri
Maurizio Scarpa
Domenica Taruscio

Temi:

- L'attività scientifica del Centro di coordinamento nazionale malattie rare per le patologie pediatriche
- Lo stato della ricerca per le malattie rare nei centri pediatrici nazionali
- La ricerca indipendente promossa da Telethon per le malattie genetiche rare
- Il ruolo dell'industria nello sviluppo dei farmaci orfani per le malattie rare
- Le reti europee per le malattie rare: censimento delle reti attive, criticità e prospettive
- Il ruolo delle Associazioni di pazienti per la ricerca sulle malattie rare pediatriche

Malattie emato-oncologiche pediatriche

A cura della Società Italiana di Ematologia e Oncologia Pediatrica - AIEOP
(Presidente: *Franca Fagioli*)

Franca Fagioli
Silverio Perrotta
Carmelo Rizzari
Marco Zecca

Pubblicazioni

Malattie oncoematologiche pediatriche: pubblicazioni dei gruppi italiani in ambito di tumori rari pediatrici

Marco Zecca
Oncoematologia Pediatrica, Fondazione Policlinico San Matteo, Pavia; Associazione Italiana di Oncoematologia Pediatrica

La Società Italiana di Ricerca Pediatrica ha avviato un'attività di *survey* relativa ad aspetti che riguardano il networking, la conduzione di studi clinici e la ricerca in ambito di malattie rare pediatriche. Scopo di questo documento è di illustrare l'attività *in progress* della creazione di un database che raccoglie i contributi pubblicativi dei centri pediatrici italiani che operano nel campo oncoematologico pediatrico.

Per quanto concerne l'attività pubblicativa in ambito di neoplasie onco-ematologiche, i dati inseriti nel database sono stati ottenuti attraverso una ricerca sulla piattaforma PubMed, inserendo quali parole chiave pediatric hemato oncology and Italy, e selezionando le pubblicazioni inerenti a patologia tumorale con primo/ultimo nome o corresponding author un autore afferente ad un centro italiano. La ricerca è stata chiusa al 31 maggio 2017.

Gli inserimenti nel database sono stati 262, distribuiti nelle seguenti categorie: 93 pubblicazioni focalizzati su neoplasie ematologiche, 123 pubblicazioni riguardanti tumori solidi e 46 lavori di tema trasversale alle patologie neoplastiche (i.e. epidemiologia, fattori di rischio, infettivologia).

Considerando le singole patologie, le pubblicazioni più numerose sono state osservate nell'ambito delle leucemie (75 pubblicazioni), seguite dalla produzione nell'ambito del neuroblastoma (26 pubblicazioni), dei sarcomi dei tessuti molli (24 pubblicazioni), dei linfomi (16 pubblicazioni) e dei tumori del sistema nervoso centrale (15 pubblicazioni). Per quanto concerne la qualità delle pubblicazioni, l'impact factor totale è di 1060 punti, con un impact factor mediano di 3,2 punti (range 0,6-26,50).

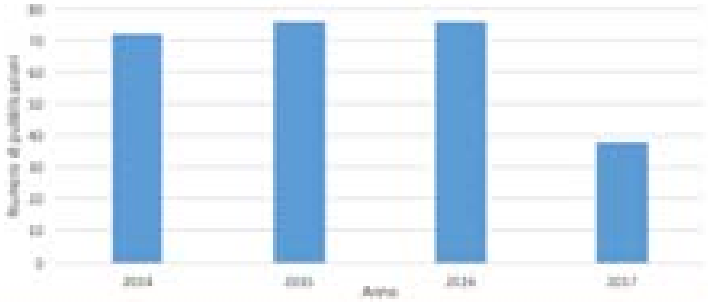
Sul totale delle registrazioni, 39 (15%) erano pubblicazioni dell'Associazione Italiana di Oncoematologia Pediatrica (pubblicazioni cooperative/linee guida dei Gruppi di Lavoro AIEOP; ricerca sulla piattaforma PubMed con parole chiave AIEOP nel titolo o nell'elenco autori). L'impact factor mediano di queste pubblicazioni è di 4,46 punti, superiore alla media dei lavori censiti all'interno del database.

Circa il 50% delle pubblicazioni incluse nel database sono il risultato di lavori cooperativi sia italiani che internazionali, a testimonianza della notevole attività collaborativa dei network nazionali ed europei oncoematologici pediatrici.

Lo stato dell'arte della produzione scientifica

Risultati

- Identificate 262 pubblicazioni.

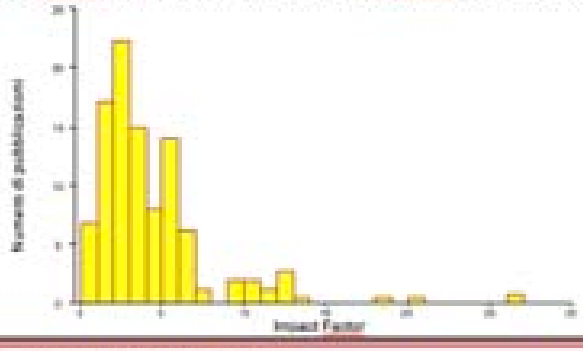


Lo stato dell'arte della produzione scientifica

Risultati

Impact factor totale = 1.060 punti

Impact factor mediano = 3,2 punti (range 0,6 - 26,50).

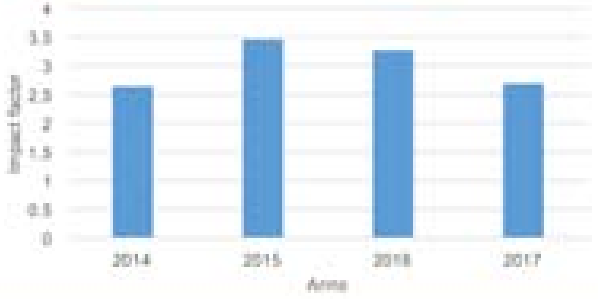


Lo stato dell'arte della produzione scientifica

Risultati

Impact factor totale = 1.060 punti

Impact factor mediano = 3,2 punti (range 0,6 - 26,50).



Lo stato dell'arte della produzione scientifica

Disease	Count	%	Cumulative count
Leukemia	75	28,63%	75
Neuroblastic tumors	26	9,92%	101
Lymphoma	16	6,11%	117
Rhabdomyosarcoma	15	5,73%	132
Other soft tissue sarcomas	9	3,44%	141
Glioblastoma	6	2,29%	147
Medulloblastoma	6	2,29%	153
Osteosarcoma	5	1,91%	158
Glioma	4	1,53%	162
Adrenocortical tumors	3	1,15%	165
Retinoblastoma	3	1,15%	168
Thyroid cancer	3	1,15%	171
Urothelial carcinoma	3	1,15%	174
Wilms tumor	3	1,15%	177
Ependymoma	2	0,76%	179
Ewing sarcoma	2	0,76%	181
Myeloproliferative disorder	2	0,76%	183
Other	33	12,60%	216
Neoplasia, generici	46	17,56%	262

Pubblicazioni: Malattie oncoematologiche pediatriche

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Adrenocortical tumors	Cerquetti L, Sampaoli C, De Salvo M, Bucci B, Argese N, Chimento A, Vottari S, Marchese R, Pezzi V, Toscano V, Stigliano A	C-MYC modulation induces responsiveness to paclitaxel in adrenocortical cancer cell lines	Int J Oncol. 46(5):2231-40.	2015	3,03	
Adrenocortical tumors	Dall'Igna P, Virgone C, De Salvo G, Bertorelle R, Indolfi P, De Paoli A, Buffa P, Conte M, Esposito G, Inserra A, Candiotto C, D'Onofrio V, Boldrini R, Ferrari A, Bisogno G, Alaggio R, Cecchetto G	Adrenocortical tumors in Italian children: Analysis of clinical characteristics and P53 status. Data from the national registries	J Pediatr Surg. 49(9):1367-71.	2014	1,31	
Adrenocortical tumors	Giovannoni I, Boldrini R, Benedetti MC, Inserra A, De Pasquale MD, Francalanci P	Pediatric adrenocortical neoplasms: immunohistochemical expression of p57 identifies loss of heterozygosity and abnormal imprinting of the 11p15.5	Pediatr Res. 81(3):468-472.	2017	2,76	
Ependymoma	Massimino M, Miceli R, Giangaspero F, Boschetti L, Modena P, Antonelli M, Ferroli P, Bertin D, Pecori E, Valentini L, Biassoni V, Garrè ML, Schiavello E, Sardi I, Cama A, Viscardi E, Scarzello G, Scoccianti S, Mascarin M, Quaglietta L, Cinalli G, Diletto B, Genitori L, Peretta P, Mussano A, Buccoliero A, Calareso G, Barra S, Mastronuzzi A, Giussani C, Marras CE, Balter R, Bertolini P, Giombelli E, La Spina M, Buttarelli FR, Pollo B, Gandola L	Final results of the second prospective AIEOP protocol for pediatric intracranial ependymoma	Neuro Oncol. 18(10):1451-60.	2016	7,37	AIEOP

Ependymoma	Roma D, Palma P, Capolino R, Figà-Talamanca L, Diomedi-Camassei F, Lepri FR, Digilio MC, Marras CE, Messina R, Carai A, Randi F, Mastronuzzi A	Spinal ependymoma in a patient with Kabuki syndrome: A case report	BMC Med Genet. 16:80.	2015	2,08	
Glioblastoma	Cacchione A, Mastronuzzi A, Cefalo MG, Colafati GS, Diomedi-Camassei F, Rizzi M, De Benedictis A, Carai A	Pediatric spinal glioblastoma of the conus medullaris: a case report of long survival	Chin J Cancer. 35:44.	2016	2,81	
Glioblastoma	Del Fattore A, Luciano R, Saracino R, Battafarano G, Rizzo C, Pascucci L, Alessandri G, Pessina A, Perrotta A, Fierabracci A, Muraca M	Differential effects of extracellular vesicles secreted by mesenchymal stem cells from different sources on glioblastoma cells	Expert Opin Biol Ther. 15(4):495-504.	2015	3,43	
Glioblastoma	Luciano R, Battafarano G, Saracino R, Rossi M, Perrotta A, Manco M, Muraca M, Del Fattore A	New perspectives in glioblastoma: Nanoparticles-based approaches	Curr Cancer Drug Targets. 17(3):203-220.	2016	3,71	
Glioblastoma	Tomaselli S, Galeano F, Alon S, Raho S, Galardi S, Polito VA, Presutti C, Vincenti S, Eisenberg E, Locatelli F, Gallo A	Modulation of microRNA editing, expression and processing by ADAR2 deaminase in glioblastoma	Genome Biol. 13;16:5.	2015	10,81	
Glioblastoma	Pistollato F, Bremer-Hoffmann S, Basso G, Cano SS, Elio I, Vergara MM, Giampieri F, Battino M.	Targeting Glioblastoma with the Use of Phytocompounds and Nanoparticles.	Target Oncol. 11(1):1-16.	2016	3,20	
Glioblastoma	Simeone P, Trerotola M, Urbanella A, Lattanzio R, Ciavardelli D, Di Giuseppe F, Eleuterio E, Sulpizio M, Eusebi V, Pession A, Piantelli M, Alberti S.	A unique four-hub protein cluster associates to glioblastoma progression.	PLoS One. 2014 Jul 22;9(7):e103030.	2014	3,23	
Glioma	Del Bufalo F, Carai A, Figa-Talamanca L, Bianco G, Cacchione A, Locatelli F, Ferretti E, Mastronuzzi A	Response of recurrent BRAFV600E mutated ganglioglioma to Vemurafenib as single agent	J Transl Med. 12:356.	2014	3,99	
Glioma	Abed E, Piccardi M, Rizzo D, Chiaretti A, Ambrosio L, Petroni S, Parrilla R, Dickmann A, Riccardi R, FalsiniB	Functional loss of the inner retina in childhood optic gliomas detected by photopic negative response	Invest Ophthalmol Vis Sci. 56(4):2469-74.	2015	3,40	

Glioma	Rossi Espagnet MC, Romano A, Mancuso V, Cicone F, Napolitano A, Scaringi C, Minniti G, Bozzao A	Multiparametric evaluation of low grade gliomas at follow-up: comparison between diffusion and perfusion MR with (18)F-FDOPA PET	Br J Radiol. 89(1066):20160476.	2016	1,84	
Glioma	Vallero SG, Bertin D, Basso ME, Pittana LS, Mussano A, Fagioli F.	Diffuse intrinsic pontine glioma in children and adolescents: a single-center experience.	Childs Nerv Syst. 30(6):1061-6.	2014	1,08	
Leukaemia	Aveic S, Viola G, Accordi B, Locatelli F, Basso G, Pigazzi M	Targeting B-cell lymphoma 2-associated AthanoGene-1 (BAG-1): A novel strategy to increase drug efficacy in acute myeloid leukemia	Exp Hematol. 43(3):180-190.e6.	2015	2,81	
Leukaemia	Paganin M, Fabbri G, Conter V, Barisone E, Polato K, Cazzaniga G, Giraldi E, Fagioli F, Aricò M, Valsecchi MG, Basso G	Postinduction minimal residual disease monitoring by polymerase chain reaction in children with acute lymphoblastic leukemia	J Clin Oncol; 32(31):3553-8.	2014	18.42	AIEOP
Leukaemia	Comoli P, Basso S, Riva G, Barozzi P, Guido I, Gurrado A, Quartuccio G, Rubert L, Lagreca I, Vallerini D, Forghieri F, Morselli M, Bresciani P, Cuoghi A, Paolini A, Colaci E, Marasca R, Cuneo A, Iughetti L, Trenti T, Narni F, Foà R, Zecca M, Luppi M, Potenza L.	BCR-ABL-specific T-cell therapy in Ph+ ALL patients on tyrosine-kinase inhibitors	Blood. 2017; 129(5):582-586.	2017	11.84	
Leukaemia	Dworzak MN, Buldini B, Gaipa G, Ratei R, Hrusak O, Luria D, Rosenthal E, Bourquin JP, Sartor M, Schumich A, Karawajew L, Mejstrikova E, Maglia O, Mann G, Ludwig WD, Biondi A, Schrappe M, Basso G; International-BFM-FLOW-network.	AIEOP-BFM consensus guidelines 2016 for flow cytometric immunophenotyping of Pediatric acute lymphoblastic leukemia	Cytometry B Clin Cytom. Feb 10. doi: 10.1002/cyto.b.21518	2017	2.47	AIEOP

Leukaemia	Buldini B, Rizzati F, Masetti R, Fagioli F, Menna G, Micalizzi C, Putti MC, Rizzari C, Santoro N, Zecca M, Disarò S, Rondelli R, Merli P, Pigazzi M, Pession A, Locatelli F, Basso G.	Prognostic significance of flow-cytometry evaluation of minimal residual disease in children with acute myeloid leukaemia treated according to the AIEOP-AML 2002/01 study protocol.	Br J Haematol. Apr;177(1):116-126.	2017	5,81	AIEOP
Leukaemia	Vendramini E, Giordan M, Giarin E, Michielotto B, Fazio G, Cazzaniga G, Biondi A, Silvestri D, Valsecchi MG, Muckenthaler MU, Kulozik AE, Gattei V, Izraeli S, Basso G, Te Kronnie G.	High expression of miR-125b-2 and SNORD116 noncoding RNA clusters characterize ERG-related B cell precursor acute lymphoblastic leukemia.	Oncotarget. Mar 21. doi: 10.18632/ oncotarget.16392	2017	5,01	
Leukaemia	Quarello P, Fagioli F, Basso G, Putti MC, Berger M, Luciani M, Rizzari C, Menna G, Masetti R, Locatelli F	Outcome of children with acute myeloid leukaemia (AML) experiencing primary induction failure in the AIEOP AML 2002/01 clinical trial	Br J Haematol. 171(4):566-73.	2015	4,97	AIEOP
Leukemia	Aveic, S, Viola, G, Accordi, B, Micalizzi, C, Santoro, N, Masetti, R, Locatelli Franco, Basso, G, Pigazzi, M.	Targeting BAG-1: A novel strategy to increase drug efficacy in acute myeloid leukemia	Exp Hematol. 43(3):180-190.e6.	2015	2,48	
Leukemia	Bisio V, Zampini M, Tregnago C, Manara E, Salsi V, Di Meglio A, Masetti R, Togni M, Di Giacomo D, Minuzzo S, Leszl A, Zappavigna V, Rondelli R, Mecucci C, Pession A, Locatelli F, Basso G, Pigazzi M	NUP98-fusion transcripts characterize different biological entities within acute myeloid leukemia: a report from the AIEOP-AML group	Leukemia. 31(4):974-977.	2016	12,10	AIEOP
Leukemia	Bresolin S, De Filippi P, Vendemini F, D'Alia M, Zecca M, Meyer LH, Danesino C, Locatelli F, Masetti R, Basso G, Te Kronnie G	Mutations of SETBP1 and JAK3 in juvenile myelomonocytic leukemia: a report from the italian AIEOP study group	Oncotarget;7(20):28914-9.	2016	5,01	AIEOP

Leukemia	Buontempo F, Orsini E, Lonetti A, Cappellini A, Bertaina A, Locatelli F, Neri LM, McCubrey JA, Martelli AM	Synergistic cytotoxic effects of bortezomib and CK2 inhibitor CX-4945 in acute lymphoblastic leukemia: turning off the prosurvival ER chaperone BIP/Grp78 and turning on the pro-apoptotic NF-kappaB	Oncotarget. 7(2):1323-40.	2016	6,36	
Leukemia	Callea M, Bellacchio E, Fattori F, Bertini ES, Callea F, Cammarata-Scalisi F	Acute myeloid leukemia in a 3 years old child with cleidocranial dysplasia	Leuk Lymphoma. 57(9):2189-91.	2016	2,89	
Leukemia	Conter V, Valsecchi M, Parasole R, Putti M, Locatelli F, Barisone E, Nigro L, Santoro N, Aricò M, Ziino O, Pession A, Testi A, Micalizzi C, Casale F, Zecca M, Casazza G, Tamaro P, La Barba G, Notarangelo L, Silvestri D, Colombini A, Rizzari C, Biondi A, Masera G, Basso G.	Childhood high-risk acute lymphoblastic leukemia in first remission: Results after chemotherapy or transplant from the AIEOP ALL 2000 study	Blood. 123(10):1470-8.	2014	9,78	AIEOP
Leukemia	Conter V, Valsecchi MG, Buldini B, Parasole R, Locatelli F, Colombini A, Rizzari C, Putti MC, Barisone E, Nigro LL, Santoro N, Ziino O, Pession A, Testi AM, Micalizzi C, Casale F, Pierani P, Cesaro S, Cellini M, Silvestri D, Cazzaniga G, Biondi A, Basso G	Early T-cell precursor acute lymphoblastic leukaemia in children treated in AIEOP centres with AIEOP-BFM protocols: a retrospective analysis	Lancet Haematol. 3(2):e80-6.	2016	4,89	AIEOP
Leukemia	D'Angiò M, Valsecchi MG, Testi AM, Locatelli F, Foa R, Biondi A, Cazzaniga G	Clinical features and outcome of SIL/TAL1-positive T-cell acute lymphoblastic leukemia in children and adolescents. A 10 year experience of the AIEOP group	Haematologica. 100(1):e10-3.	2015	5,87	AIEOP

Leukemia	Decembrino N, Zecca M, Tortorano AM, Mangione F, Lallitto F, Introzzi F, Bergami E, Marone P, Tamarozzi F, Cavanna C	Acute isolated appendicitis due to Aspergillus carneus in a neutropenic child with acute myeloid leukemia	New Microbiol; 39(1):65-9	2016	1,72	
Leukemia	De Felice F, Grapulin L, Musio D, Pomponi J, DI Felice C, Iori AP, Bertaina A, Tombolini V	Treatment Complications and Long-term Outcomes of Total Body Irradiation in Patients with Acute Lymphoblastic Leukemia: A Single Institute Experience	Anticancer Res. 36(9):4859-64.	2016	1,90	
Leukemia	de Rooij JD, Masetti R, van den Heuvel-Eibrink MM, Cayuela JM, Trka J, Reinhardt D, Rasche M, Sonneveld E, Alonzo TA, Fornerod M, Zimmermann M, Pigazzi M, Pieters R, Meshinchi S, Zwaan CM, Locatelli F	Recurrent abnormalities can be used for risk group stratification in pediatric AMKL: a retrospective intergroup study	Blood. 127(26):3424-30.	2016	11,85	
Leukemia	Evangelisti C, Evangelisti C, Teti G, Chiarini F, Falconi M, Melchionda F, Pession A, Bertaina Alice, Locatelli F, McCubrey J, Beak D, Bittman R, Pyne S, Pyne N, Martelli A	Assessment of the effect of sphingosine kinase inhibitors on apoptosis,unfolded protein response and autophagy of T-cell acute lymphoblastic leukemia cells; Indications for novel therapeutics	Oncotarget. 5(17):7886-901.	2014	6,63	
Leukemia	Folgiero V, Cifaldi L, Li Pira G, Goffredo BM, Vinti L, Locatelli F	TIM-3/Gal-9 interaction induces IFNÎ³-dependent IDO1 expression in acute myeloid leukemia blast cells	J Hematol Oncol. 8:36.	2015	4,81	
Leukemia	Folgiero V, Goffredo BM, Filippini P, Masetti R, Bonanno G, Caruso R, Bertaina V, Mastronuzzi A, Gaspari S, Zecca M, Torelli G, Testi A, Pession A, Locatelli F, Rutella S	Indoleamine 2,3-dioxygenase 1 (IDO1) activity in leukemia blasts correlates with poor outcome in childhood acute myeloid leukemia	Oncotarget. 5(8):2052-64.	2014	6,63	

Leukemia	Franca R, Rebora P, Bertorello N, Fagioli F, Conter V, Biondi A, Colombini A, Micalizzi C, Zecca M, Parasole R, Petruzziello F, Basso G, Putti MC, Locatelli F, d'Adamo P, Valsecchi MG, Decorti G, Rabusin M	Pharmacogenetics and induction/consolidation therapy toxicities in acute lymphoblastic leukemia patients treated with AIEOP-BFM ALL 2000 protocol	Pharmacogenomics J. 17(1):4-10.	2017	4,23	AIEOP
Leukemia	Giona F, Putti MC, Micalizzi C, Menna G, Moleti ML, Santoro N, Iaria G, Ladogana S, Burnelli R, Consarino C, Varotto S, Tucci F, Messina C, Nanni M, Diverio D, Biondi A, Pession A, Locatelli F, Piciocchi A, Gottardi E, Saglio G, Foà R	Long-term results of high-dose imatinib in children and adolescents with chronic myeloid leukaemia in chronic phase: the Italian experience	Br J Haematol. 170(3):398-407.	2015	4,97	
Leukemia	Giona F, Saglio G, Santopietro M, Menna G, Putti MC, Micalizzi C, Iaria G, Santoro N, Ladogana S, Mura R, Burnelli R, Consarino C, Cosmi C, Moleti ML, Leszl A, Tucci F, Nanni M, Diverio D, Biondi A, Locatelli F, Foà R	Early response does not predict outcome in children and adolescents with chronic myeloid leukaemia treated with high-dose imatinib	Br J Haematol. doi: 10.1111/bjh.14458.	2016	5,81	
Leukemia	Leoncini PP, Bertaina A, Papaioannou D, Flotho C, Masetti R, Bresolin S, Menna G, Santoro N, Zecca M, Basso G, Nigita G, Veneziano D, Pagotto S, D'Ovidio K, Rota R, Dorrance A, Croce CM, Niemeyer C, Locatelli F, Garzon R	MicroRNA fingerprints in juvenile myelomonocytic leukemia (JMML) identified miR-150-5p as a tumor suppressor and potential target for treatment	Oncotarget. 7(34):55395-55408.	2016	5,01	
Leukemia	Lepore M, De Lalla C, Gundimeda R, Gsellinger H, Consonni M, Garavaglia C, Sansano S, Piccolo F, Scelfo A, Häussinger D, Montagna D, Locatelli F, Bonini C, Bondanza A, Forcina A, Li A, Ni G, Ciceri F, Jenö P, Xia C, Mori L, Dellabona P, Casorati G, de Libero G.	A novel self-lipid antigen targets human T cells against CD1c⁺ leukemias	J Exp Med. 211(7):1363-77.	2014	13,91	
Leukemia	Locatelli F and Niemeyer CM	How I treat juvenile myelomonocytic leukemia	Blood. 125(7):1083-90.	2015	10,45	

Leukemia	Locatelli F, Masetti R, Rondelli R, Zecca M, Fagioli F, Rovelli A, Messina C, Lanino E, Bertaina A, Favre C, Giorgiani G, Ripaldi M, Ziino O, Palumbo G, Pillon M, Pession A, Rutella S, Prete A.	Outcome of children with high-risk acute myeloid leukemia given autologous or allogeneic hematopoietic cell transplantation in the aieop AML-2002/01 study	Bone Marrow Transplant. 50(2):181-8.	2015	3,47	AIEOP
Leukemia	Locatelli F, Moretta F, Brescia LP, Merli P	Natural killer cells in the treatment of high-risk acute leukaemia	Semin Immunol. 26(2):173-9.	2014	6,12	
Leukemia	Franca R, Rebora P, Athanasakis E, Favretto D, Verzegnassi F, Basso G, Tommasini A, Valsecchi MG, Decorti G, Rabusin M	TNF-α SNP rs1800629 and risk of relapse in childhood acute lymphoblastic leukemia: relation to immunophenotype	Pharmacogenomics; 15(5):619-27	2014	3,22	AIEOP
Leukemia	Milani G, Rebora P, Accordi B, Galla L, Bresolin S, Cazzaniga G, Buldini B, Mura R, Ladogana S, Giraldi E, Conter V, Te Kronnie G, Valsecchi MG, Basso G	Low PKCα expression within the MRD-HR stratum defines a new subgroup of childhood T-ALL with very poor outcome	Oncotarget; 5(14):5234-45.	2014	6,36	AIEOP
Leukemia	Manara E, Baron E, Tregnago C, Aveic S, Bisio V, Bresolin S, Masetti R, Locatelli F, Basso G, Pigazzi M	MLL-AF6 fusion oncogene sequesters AF6 into the nucleus to trigger RAS activation in myeloid leukemia	Blood. 124(2):263-72.	2014	9,78	
Leukemia	Manara E, Basso G, Zampini M, Buldini B, Tregnago C, Rondelli R, Masetti R, Bisio V, Frison M, Polato K, Cazzaniga G, Menna G, Fagioli F, Merli P, Biondi A, Pession A, Locatelli F, Pigazzi M	Characterization of children with FLT3-ITD acute myeloid leukemia: a report from the AIEOP AML-2002 study group	Leukemia. 31(1):18-25.	2017	12,10	AIEOP
Leukemia	Manara E, Bisio V, Masetti R, Beqiri V, Rondelli R, Menna G, Micalizzi C, Santoro N, Locatelli F, Basso G, Pigazzi M	Core-binding factor acute myeloid leukemia in pediatric patients enrolled in the AIEOP AML 2002/01 trial: Screening and prognostic impact of c-KIT mutations	Leukemia. 28(5):1132-4.	2014	9,38	AIEOP
Leukemia	Masetti R, Castelli I, Astolfi A, Bertuccio SN, Indio V, Togni M, Belotti T, Serravalle S, Tarantino G, Zecca M, Pigazzi M, Basso G, Pession A, Locatelli F	Genomic complexity and dynamics of clonal evolution in childhood acute myeloid leukemia studied with whole-exome sequencing	Oncotarget. 7(35):56746-56757.	2016	5,01	

Leukemia	Masetti R, Rondelli R, Fagioli F, Mastronuzzi A, Pierani P, Togni M, Menna G, Pigazzi M, Putti M, Basso G, Pession A, Locatelli F	Infants with acute myeloid leukemia treated according to the Associazione Italiana di Ematologia e Oncologia Pediatrica 2002/01 protocol have an outcome comparable to that of older children	Haematologica. 99(8):e127-9.	2014	5,87	AIEOP
Leukemia	Masetti R, Togni M, Astolfi A, Pigazzi M, Indio V, Rivalta B, Manara E, Rutella S, Basso G, Pession A, Locatelli F	Whole transcriptome sequencing of a paediatric case of de novo acute myeloid leukaemia with del(5q) reveals RUNX1-USP42 and PRDM16-SKI fusion transcripts	Br J Haematol. 166(3):449-52.	2014	4,96	
Leukemia	Palmi C, Savino AM, Silvestri D, Bronzini I, Cario G, Paganin M, Buldini B, Galbiati M, Muckenthaler MU, Bugarin C, Della Mina P, Nagel S, Barisone E, Casale F, Locatelli F, Lo Nigro L, Micalizzi C, Parasole R, Pession A, Putti MC, Santoro N, Testi AM, Ziino O, Kulozik AE, Zimmermann M, Schrappe M, Villa A, Gaipa G, Basso G, Biondi A, Valsecchi MG, Stanulla M, Conter V, Te Kronnie G, Cazzaniga G	CRLF2 over-expression is a poor prognostic marker in children with high risk T-cell acute lymphoblastic leukemia	Oncotarget. 7(37):59260-59272.	2016	5,01	AIEOP
Leukemia	Parasole R, Petruzziello F, Messina C, Locatelli F, Berger M, Morello W, Menna G, Poggi V	Toxicity and efficacy of intrathecal liposomal cytarabine in children with leukemia/lymphoma relapsing in the central nervous system (CNS): a retrospective multicenter study	Leuk Lymphoma. 56(3):650-5.	2015	2,61	

Leukemia	Pigazzi M, Manara E, Buldini B, Beqiri V, Bisio V, Tregnago C, Rondelli R, Masetti R, Putti MC, Fagioli F, Rizzari C, Pession A, Locatelli F, Basso G	Minimal residual disease monitored after induction therapy by RQ-PCR can contribute to tailor treatment of patients with t(8;21) RUNX1-RUNX1T1 rearrangement	Haematologica. 100(3):e99-101.	2015	5,87	AIEOP
Leukemia	Spartà A, Bressanin D, Chiarini F, Lonetti A, Cappellini A, Evangelisti C, Evangelisti C, Melchionda F, Pession A, Bertaina A, Locatelli F, McCubrey J, Martelli A	Therapeutic targeting of Polo-like kinase-1 and Aurora kinases in T-cell acute lymphoblastic leukemia	Cell Cycle. 13(14):2237-47.	2014	5,01	
Leukemia	Cesaro S, De Filippi P, Di Meglio A, Leszl A, Donska S, Zaccaron A, Cagioni C, Galavotti R, Danesino C, Aprili F, Cugno C, te Kronnie G, Zecca M, Bresolin S	Different outcomes of allogeneic hematopoietic stem cell transplant in a pair of twins affected by juvenile myelomonocytic leukemia	Int J Hematol; 99(2):208-12	2014	1,84	
Leukemia	Testi AM, Moleti ML, Canichella M, Mohamed S, Diverio D, de Propris MS, Locatelli F, Lo Coco F, Foà R	Very late relapse in a patient with acute promyelocytic leukemia (APL) rescued with a chemotherapy-free protocol	Leuk Lymphoma. 58(4):999-1001.	2017	3,09	
Leukemia	Togni M, Masetti R, Pigazzi M, Astolfi A, Zama D, Indio V, Serravalle S, Manara E, Bisio V, Rizzari C, Basso G, Pession A, Locatelli F	Identification of the NUP98-PHF23 fusion gene in pediatric cytogenetically normal acute myeloid leukemia by whole-transcriptome sequencing	J Hematol Oncol. 8:69.	2015	4,81	AIEOP
Leukemia	Torelli G, Peragine N, Raponi S, Pagliara D, De Propris M, Vitale A, Bertaina A, Barberi W, Moretta L, Basso G, Santoni A, Guarini A, Locatelli F, Foà R	Recognition of adult and pediatric acute lymphoblastic leukemia blasts by natural killer cells	Haematologica. 99(7):1248-54.	2014	5,87	
Leukemia	Bugarin C, Sarno J, Palmi C, Savino AM, te Kronnie G, Dworzak M, Shumich A, Buldini B, Maglia O, Sala S, Bronzini I, Bourquin JP, Mejstrikova E, Hrusak O, Luria D, Basso G, Izraeli S, Biondi A, Cazzaniga G, Gaipa G; I-BFM study group.	Fine tuning of surface CRLF2 expression and its associated signaling profile in childhood B-cell precursor acute lymphoblastic leukemia.	Haematologica. 100(6):e229-32.	2015	5,87	

Leukemia	Lana T, de Lorenzo P, Bresolin S, Bronzini I, den Boer ML, Cavé H, Froňková E, Stanulla M, Zaliova M, Harrison CJ, de Groot H, Valsecchi MG, Biondi A, Basso G, Cazzaniga G, te Kronnie G.	Refinement of IKZF1 status in pediatric Philadelphia-positive acute lymphoblastic leukemia.	Leukemia. 29(10):2107-10.	2015	12,10	
Leukemia	Cazzaniga V, Bugarin C, Bardini M, Giordan M, te Kronnie G, Basso G, Biondi A, Fazio G, Cazzaniga G.	LCK over-expression drives STAT5 oncogenic signaling in PAX5 translocated BCP-ALL patients.	Oncotarget. 6(3):1569-81.	2015	5,01	
Leukemia	Fazio G, Daniele G, Cazzaniga V, Impera L, Severgnini M, Iacobucci I, Galbiati M, Leszl A, Cifola I, De Bellis G, Bresciani P, Martinelli G, Basso G, Biondi A, Storlazzi CT, Cazzaniga G.	Three novel fusion transcripts of the paired box 5 gene in B-cell precursor acute lymphoblastic leukemia.	Haematologica. 100(1):e14-7.	2015	0,27	
Leukemia	Dall'Acqua S, Linardi MA, Bortolozzi R, Clauser M, Marzocchini S, Maggi F, Nicoletti M, Innocenti G, Basso G, Viola G.	Natural daucane esters induces apoptosis in leukaemic cells through ROS production.	Phytochemistry. 108:147-56.	2014	2,55	
Leukemia	La Starza R, Borga C, Barba G, Pierini V, Schwab C, Matteucci C, Lema Fernandez AG, Leszl A, Cazzaniga G, Chiaretti S, Basso G, Harrison CJ, Te Kronnie G, Mecucci C.	Genetic profile of T-cell acute lymphoblastic leukemias with MYC translocations.	Blood. 124(24):3577-82.	2014	10,45	
Leukemia	Bortolozzi R, Viola G, Porcù E, Consolaro F, Marzano C, Pellei M, Gandin V, Basso G.	A novel copper(I) complex induces ER-stress-mediated apoptosis and sensitizes B-acute lymphoblastic leukemia cells to chemotherapeutic agents.	Oncotarget. 5(15):5978-91.	2014	6,35	
Leukemia	Cavazzini F, Campioni D, Ferrari L, Buldini B, Bardi MA, Michielotto B, Lazzari MC, Ongari M, Dabusti M, Daghia G, Sofritti O, Basso G, Lanza F, Cuneo A.	Expression of the immunoglobulin superfamily cell membrane adhesion molecule Cd146 in acute leukemia.	Cytometry B Clin Cytom. 90(3):247-56.	2016	2,82	

Leukemia	Consolaro F, Ghaem-Maghami S, Bortolozzi R, Zona S, Khongkow M, Basso G, Viola G, Lam EW.	FOXO3a and Posttranslational Modifications Mediate Glucocorticoid Sensitivity in B-ALL	Mol Cancer Res. 13(12):1578-90.	2015	4,51	
Leukemia	Consolaro F, Basso G, Ghaem-Magami S, Lam EW, Viola G.	FOXM1 is overexpressed in B-acute lymphoblastic leukemia (B-ALL) and its inhibition sensitizes B-ALL cells to chemotherapeutic drugs.	Int J Oncol. 47(4):1230-40.	2015	3,02	
Leukemia	Germano G, Morello G, Aveic S, Pinazza M, Minuzzo S, Frasson C, Persano L, Bonvini P, Viola G, Bresolin S, Tregnago C, Paganin M, Pigazzi M, Indraccolo S, Basso G.	ZNF521 sustains the differentiation block in MLL-rearranged acute myeloid leukemia.	Oncotarget. 8(16):26129-26141.	2017	5,01	
Leukemia	Serafin V, Lissandron V, Buldini B, Bresolin S, Paganin M, Grillo F, Andriano N, Palmi C, Cazzaniga G, Marmiroli S, Conter V, Basso G, Accordi B.	Phosphoproteomic analysis reveals hyperactivation of mTOR/STAT3 and LCK/Calcineurin axes in pediatric early T-cell precursor ALL.	Leukemia. 31(4):1007-1011.	2017	12,10	
Leukemia	Frezzato F, Accordi B, Trimarco V, Gattazzo C, Martini V, Milani G, Bresolin S, Severin F, Visentin A, Basso G, Semenzato G, Trentin L.	Profiling B cell chronic lymphocytic leukemia by reverse phase protein array: Focus on apoptotic proteins.	J Leukoc Biol. 100(5):1061-1070.	2016	4,17	
Leukemia	Magnani C, Ranucci A, Badaloni C, Cesaroni G, Ferrante D, Miligi L, Mattioli S, Rondelli R, Bisanti L, Zambon P, Cannizzaro S, Michelozzi P, Cocco P, Celentano E, Assennato G, Merlo DF, Mosciatti P, Minelli L, Cuttini M, Torregrossa MV, Lagorio S, Haupt R, Forastiere F; SETIL Working Group.	Road Traffic Pollution and Childhood Leukemia: A Nationwide Case-control Study in Italy.	Arch Med Res. 47(8):694-705.	2016	2,21	
Leukemia	Paganin M, Buldini B, Germano G, Seganfreddo E, Meglio Ad, Magrin E, Grillo F, Pigazzi M, Rizzari C, Cazzaniga G, Khiabanian H, Palomero T, Rabadan R, Ferrando AA, Basso G.	A Case of T-cell Acute Lymphoblastic Leukemia Relapsed As Myeloid Acute Leukemia.	Pediatr Blood Cancer. 63(9):1660-3.	2016	2,63	

Leukemia	Cazzaniga G, Bisanti L, Randi G, Deandrea S, Bungaro S, Pregliasco F, Perotti D, Spreafico F, Masera G, Valsecchi MG, Biondi A, Greaves M.	Possible role of pandemic AH1N1 swine flu virus in a childhood leukemia cluster	Leukemia Apr 27. doi: 10.1038/leu.2017.127	2017	12,10	
Leukemia	Bruzzi P, Predieri B, Corrias A, Marsciani A, Street ME, Rossidivita A, Paolucci P, Iughetti L	Final height and body mass index in adult survivors of childhood acute lymphoblastic leukemia treated without cranial radiotherapy: a retrospective longitudinal multicenter Italian study	BMC Pediatr. 2014 Sep 22;14:236	2014	1,90	
Leukemia	Cianci P, Fazio G, Casagranda S, Spinelli M, Rizzari C, Cazzaniga G, Selicorni A	Acute myeloid leukemia in Baraitser-Winter cerebrofrontofacial syndrome	Am J Med Genet A; 173(2):546-549	2017	2,08	
Leukemia	Tosello V, Bordin F, Yu J, Agnusdei V, Indraccolo S, Basso G, Amadori A, Piovan E.	Calcineurin and GSK-3 inhibition sensitizes T-cell acute lymphoblastic leukemia cells to apoptosis through X-linked inhibitor of apoptosis protein degradation.	Leukemia. 30(4):812-22.	2016	12,10	
Leukemia	Astolfi A, Vendemini F, Urbini M, Melchionda F, Masetti R, Franzoni M, Libri V, Serravalle S, Togni M, Paone G, Montemurro L, Bressanin D, Chiarini F, Martelli AM, Tonelli R, Pession A.	MYCN is a novel oncogenic target in pediatric T-cell acute lymphoblastic leukemia.	Oncotarget. 5(1):120-30.	2014	6,36	
Leukemia	Lonetti A, Antunes IL, Chiarini F, Orsini E, Buontempo F, Ricci F, Tazzari PL, Pagliaro P, Melchionda F, Pession A, Bertaina A, Locatelli F, McCubrey JA, Barata JT, Martelli AM.	Activity of the pan-class I phosphoinositide 3-kinase inhibitor NVP-BKM120 in T-cell acute lymphoblastic leukemia.	Leukemia. 28(6):1196-206.	2014	10,43	
Leukemia	Buontempo F, Orsini E, Martins LR, Antunes I, Lonetti A, Chiarini F, Tabellini G, Evangelisti C, Evangelisti C, Melchionda F, Pession A, Bertaina A, Locatelli F, McCubrey JA, Cappellini A, Barata JT, Martelli AM.	Cytotoxic activity of the casein kinase 2 inhibitor CX-4945 against T-cell acute lymphoblastic leukemia: targeting the unfolded protein response signaling.	Leukemia. 28(3):543-53.	2014	10,43	

Leukemia	Serravalle S, Bertuccio SN, Astolfi A, Melchionda F, Pession A.	Synergistic Cytotoxic Effect of L-Asparaginase Combined with Decitabine as a Demethylating Agent in Pediatric T-ALL, with Specific Epigenetic Signature.	Biomed Res Int. 2016:1985750	2016	2,48	
Leukemia	Lonetti A, Cappellini A, Bertaina A, Locatelli F, Pession A, Buontempo F, Evangelisti C, Evangelisti C, Orsini E, Zambonin L, Neri LM, Martelli AM, Chiarini F.	Improving nelarabine efficacy in T cell acute lymphoblastic leukemia by targeting aberrant PI3K/AKT/mTOR signaling pathway.	J Hematol Oncol. 9(1):114.	2016	6,62	
Leukemia	Masetti R, Bertuccio SN, Astolfi A, Chiarini F, Lonetti A, Indio V, De Luca M, Bandini J, Serravalle S, Franzoni M, Pigazzi M, Martelli AM, Basso G, Locatelli F, Pession A.	Hh/Gli antagonist in acute myeloid leukemia with CBFA2T3-GLIS2 fusion gene.	J Hematol Oncol. 10(1):26.	2017	6,26	
Leukemia	Cistaro A, Saglio F, Asaftei S, Fania P, Berger M, Fagioli F.	The role of 18F-FDG PET/CT in pediatric lymph-node acute lymphoblastic leukemia involvement.	Radiol Case Rep. 6(4):503.	2015	0,04	
Leukemia	Rizzari C	Still trying to pick the best asparaginase preparation	Lancet Oncol; 16(16):1580-1	2015	26,50	
Leukemia	Pillozzi S, Masselli M, Gasparoli L, D'Amico M, Polletta L, Veltroni M, Favre C, Basso G, Becchetti A, Arcangeli A	Macrolide antibiotics exert antileukemic effects by modulating the autophagic flux through inhibition of hERG1 potassium channels	Blood Cancer J; 6:e423	2016	4,41	
Leukemia	Faraci M, Matthes-Martin S, Lanino E, Morreale G, Ferretti M, Giardino S, Micalizzi C, Balduzzi A.	Two pregnancies shortly after transplantation with reduced intensity conditioning in chronic myeloid leukemia.	Pediatr Transplant. 20(1):158-61.	2016	1,28	

Leukemia/Lymphoma	Berger M, Lanino E, Cesaro S, Zecca M, Vassallo E, Faraci M, De Bortoli M, Barat V, Prete A, Fagioli F	Feasibility and Outcome of Haploidentical Hematopoietic Stem Cell Transplantation with Post-Transplant High-Dose Cyclophosphamide for Children and Adolescents with Hematologic Malignancies: An AIEOP-GITMO Retrospective Multicenter Study	Biol Blood Marrow Transplant; 22(5):902-9.	2016	4,70	AIEOP
Lymphoma	Cambiaso P, Bottaro G, Cianfarani S, Toma P, Vito RD, Cappa M	An Incidental Finding of Bilateral Adrenal Lymphoma	Am J Med Sci. 352(1):80.	2016	1,58	
Lymphoma	Attarbaschi A, Carraro E, Abla O, Barzilai-Birenboim S, Bomken S, Brugieres L, Bubanska E, Burkhardt B, Chiang AK, Csoka M, Fedorova A, Jazbec J, Kabickova E, Krenova Z, Lazic J, Loeffen J, Mann G, Niggli F, Miakova N, Osumi T, Ronceray L, Uyttebroeck A, Williams D, Woessmann W, Wrobel G, Pillon M; European Intergroup for Childhood Non-Hodgkin Lymphoma (EICNHL) and the International Berlin-Frankfur t-Münster (i-BFM) Study Group	Non-Hodgkin lymphoma and pre-existing conditions: spectrum, clinical characteristics and outcome in 213 children and adolescents	Haematologica;101(12):1581-1591	2016	6,67	
Lymphoma	Pillon M, Aricò M, Mussolin L, Mainardi C, Giraldi E, Garaventa A, Lombardi A, Santoro N, Carraro E, d'Amore ES, Rosolen A; Italian Association of Pediatric Hematology, Oncology AIEOP Non-Hodgkin Lymphoma Working Group	Mediastinal Burkitt lymphoma in childhood.	Pediatr Blood Cancer; 61(11):2127-8.	2014	2,63	AIEOP
Lymphoma	Aricò M, Mussolin L, Carraro E, Buffardi S, Santoro N, D'Angelo P, Lombardi A, Pierani P, Giraldi E, Mura R, Sala A, Garaventa A, Tondo A, Piglione M, Lo Nigro L, Cesaro S, Perruccio K, Rosolen	Non-Hodgkin lymphoma in children with an associated inherited condition: A retrospective analysis of the Associazione Italiana Ematologia Oncologia Pediatrica	Pediatr Blood Cancer; 62(10):1782-9.	2015	2,39	AIEOP

	A, Basso G, Pillon M	(AIEOP).				
Lymphoma	Farruggia P, Puccio G, Sala A, Todesco A, Buffardi S, Garaventa A, Bottigliero G, Bianchi M, Zecca M, Locatelli F, Pession A, Pillon M, Favre C, D'Amico S, Provenzi M, Trizzino A, Zanazzo GA, Sau A, Santoro N, Murgia G, Casini T, Mascarin M, Burnelli R; AIEOP Italian Association of Pediatric Hematology and Oncology and Hodgkin Lymphoma Working Group	The prognostic value of biological markers in paediatric Hodgkin lymphoma	Eur J Cancer. 52:33-40.	2016	5,42	AIEOP
Lymphoma	Farruggia P, Puccio G, Sala A, Todesco A, Terenziani M, Mura R, D'Amico S, Casini T, Mosa C, Pillon M, Boaro MP, Bottigliero G, Burnelli R, Consarino C, Fedeli F, Mascarin M, Perruccio K, Schiavello E, Trizzino A, Ficola U, Garaventa A, Rossello M	Abdomen/pelvis computed tomography in staging of pediatric Hodgkin Lymphoma: is it always necessary?	Cancer Med. 2016 Sep;5(9):2359-67.	2016	3,36	
Lymphoma	Mussolin L, Buldini B, Lovisa F, Carraro E, Disarò S, Lo Nigro L, d'Amore ES, Pillon M, Basso G	Detection and role of minimal disseminated disease in children with lymphoblastic lymphoma: The AIEOP experience	Pediatr Blood Cancer; 62(11):1906-13.	2015	2,39	AIEOP
Lymphoma	Pillon M, Aricò M, Mussolin L, Carraro E, Conter V, Sala A, Buffardi S, Garaventa A, D'Angelo P, Lo Nigro L, Santoro N, Piglione M, Lombardi A, Porta F, Cesaro S, Moleti ML, Casale F, Mura R, d'Amore ES, Basso G, Rosolen A	Long-term results of the AIEOP LNH-97 protocol for childhood lymphoblastic lymphoma	Pediatr Blood Cancer. 62(8):1388-94.	2015	2,39	AIEOP
Lymphoma	Pillon M, Arico M, Mussolin L, Lombardi A, Carraro E, D'Amore ES, Rosolen A	Mediastinal Burkitt lymphoma in childhood	Pediatr Blood Cancer. 61(11):2127-8.	2014	2,56	AIEOP

Lymphoma	Pillon M, Mussolin L, Carraro E, Conter V, Aricò M, Vinti L, Garaventa A, Piglione M, Buffardi S, Sala A, Santoro N, Lo Nigro L, Mura R, Tondo A, Casale F, Farruggia P, Pierani P, Cesaro S, d'Amore ES, Basso G	Detection of prognostic factors in children and adolescents with Burkitt and Diffuse Large B-Cell Lymphoma treated with the AIEOP LNH-97 protocol	Br J Haematol. 175(3):467-475.	2016	5,81	AIEOP
Lymphoma	Primerano S, Burnelli R, Carraro E, Pillon M, Elia C, Farruggia P, Sala A, Vinti L, Buffardi S, Basso G, Mascarin M, Mussolin L	Kinetics of Circulating Plasma Cell-Free DNA in Paediatric Classical Hodgkin Lymphoma	J Cancer. 7(4):364-6.	2016	3,61	
Lymphoma	Lovisa F, Cozza G, Cristiani A, Cuzzolin A, Albiero A, Mussolin L, Pillon M, Moro S, Basso G, Rosolen A, Bonvini P.	ALK kinase domain mutations in primary anaplastic large cell lymphoma: consequences on NPM-ALK activity and sensitivity to tyrosine kinase inhibitors.	PLoS One. 10(4):e0121378.	2015	3,54	
Lymphoma	Pomari E, Basso G, Bresolin S, Pillon M, Carraro E, d'Amore ES, Viola G, Frasson C, Basso K, Bonvini P, Mussolin L.	NPM-ALK expression levels identify two distinct subtypes of paediatric anaplastic large cell lymphoma.	Leukemia. 31(2):498-501.	2017	12,10	
Lymphoma	Ferrari C, Niccoli Asabella A, Merenda N, Altini C, Fanelli M, Muggeo P, De Leonardis F, Perillo T, Santoro N, Rubini G	Pediatric Hodgkin Lymphoma: Predictive value of interim 18F-FDG PET/CT in therapy response assessment	Medicine (Baltimore). Feb;96(5):e5973.	2017	1,20	
Lymphoma	Ciambotti B, Mussolin L, d'Amore ES, Pillon M, Sieni E, Coniglio ML, Ros MD, Cetica V, Aricò M, Rosolen A.	Monoallelic mutations of the perforin gene may represent a predisposing factor to childhood anaplastic large cell lymphoma.	J Pediatr Hematol Oncol; 36(6):e359-65	2014	0,38	
Lymphoma	Mediani L, Gibellini F, Bertacchini J, Frasson C, Bosco R, Accordi B, Basso G, Bonora M, Calabrò ML, Mattiolo A, Sgarbi G, Baracca A, Pinton P, Riva G, Rampazzo E, Petrizza L, Prodi L, Milani D, Luppi M, Potenza L, De Pol A, Cocco L, Capitani S, Marmiroli S.	Reversal of the glycolytic phenotype of primary effusion lymphoma cells by combined targeting of cellular metabolism and PI3K/Akt/ mTOR signaling.	Oncotarget. 7(5):5521-37.	2016	5,01	

Medulloblastoma	Massimino M, Biassoni V, Gandola L, Garrè ML, Gatta G, Giangaspero F, Poggi G, Rutkowski S	Childhood medulloblastoma	Crit Rev Oncol Hematol; 105:35-51	2016	5,04	
Medulloblastoma	Frasson C, Rampazzo E, Accordi B, Beggio G, Pistollato F, Basso G, Persano L.	Inhibition of PI3K Signalling Selectively Affects Medulloblastoma Cancer Stem Cells.	Biomed Res Int. 2015:973912.	2015	2,48	
Medulloblastoma	Catanzaro G, Besharat ZM, Garg N, Ronci M, Pieroni L, Miele E, Mastronuzzi A, Carai A, Alfano V, Po A, Screpanti I, Locatelli F, Urbani A, Ferretti E	MicroRNAs-Proteomic Networks Characterizing Human Medulloblastoma-SLCs	Stem Cells Int. 2016:2683042.	2016	3,69	
Medulloblastoma	Di Giannatale A, Carai A, Cacchione A, Marrazzo A, Dell'Anna VA, Colafati GS, Diomedi-Camassei F, Miele E, Po A, Ferretti E, Locatelli F, Mastronuzzi A	Anomalous vascularization in a Wnt medulloblastoma: a case report	BMC Neurol. 16:103.	2016	1,96	
Medulloblastoma	Folgiero V, Miele E, Carai A, Ferretti E, Alfano V, Po A, Bertaina V, Goffredo BM, Benedetti MC, Camassei FD, Cacchione A, Locatelli F, Mastronuzzi A	IDO1 involvement in mTOR pathway: A molecular mechanism of resistance to mTOR targeting in medulloblastoma	Oncotarget. 7(33):52900-52911.	2016	5,01	
Medulloblastoma	Mastronuzzi A, Miele E, Po A, Antonelli M, Buttarelli F, Colafati G, Del Bufalo F, Faedda R, Spinelli G, Carai A, Giangaspero F, Gulino A, Locatelli F, Ferretti E	Large cell anaplastic medulloblastoma metastatic to the scalp: Tumor and derived stem-like cells features	BMC Cancer. 14:262.	2014	3,32	
Medulloepithelioma	De Pasquale MD, De Ioris MA, Gallo A, Mastronuzzi A, Crocoli A, Cozza R, Boldrini R	Peripheral medulloepithelioma: A rare tumor with a potential target therapy	J Transl Med. 12:49.	2014	3,99	
Meningiomas	Felicetti F, Fortunati N, Garbossa D, Biasin E, Rudà R, Daniele D, Arvat E, Corrias A, Fagioli F, Brignardello E.	Meningiomas after cranial radiotherapy for childhood cancer: a single institution experience.	J Cancer Res Clin Oncol. 141(7):1277-82.	2015	3,14	

Myeloproliferative disorder	Avanzini M, Bernardo ME, Novara F, Mantelli M, Poletto V, Villani L, Lenta E, Ingo D, Achille V, Bonetti E, Massa M, Campanelli R, Fois G, Catarsi P, Gale R, Moretta A, Aronica A, Maccario R, Acquafredda G, Lisini D, Zecca M, Zuffardi O, Locatelli F, Barosi G, Rosti V	Functional and genetic aberrations of in vitro-cultured marrow-derived mesenchymal stromal cells of patients with classical Philadelphia-negative myeloproliferative neoplasms	Leukemia. 28(8):1742-5.	2014	9,38	
Myeloproliferative disorder	Romano M, Della Porta MG, Gallì A, Panini N, Licandro SA, Bello E, Craparotta I, Rosti V, Bonetti E, Tancredi R, Rossi M, Mannarino L, Marchini S, Porcu L, Galmarini CM, Zambelli A, Zecca M, Locatelli F, Cazzola M, Biondi A, Rambaldi A, Allavena P, Erba E, D'Incalci M	Antitumour activity of trabectedin in myelodysplastic/myeloproliferative neoplasms.	Br J Cancer; 116(3):335-343.	2017	5,57	
Neoplasia, general	Principi N, Preti V, Gaspari S, Colombini A, Zecca M, Terranova L, Cefalo MG, Ierardi V, Pelucchi C, Esposito S.	Streptococcus pneumoniae pharyngeal colonization in school-age children and adolescents with cancer.	Hum Vaccin Immunother;12:301-7.	2016	2,14	
Neoplasia, general	Beneventi F, Locatelli E, Giorgiani G, Zecca M, Locatelli F, Cavagnoli C, Simonetta M, Bariselli S, Negri B, Spinillo A	Gonadal and uterine function in female survivors treated by chemotherapy, radiotherapy, and/or bone marrow transplantation for childhood malignant and non-malignant diseases	BJOG;121(7):856-65	2014	3,44	
Neoplasia, general	Beneventi F, Locatelli E, Giorgiani G, Zecca M, Mina T, Simonetta M, Cavagnoli C, Albanese M, Spinillo A.	Adolescent and adult uterine volume and uterine artery Doppler blood flow among subjects treated with bone marrow transplantation or chemotherapy in pediatric age: a case-control study.	Fertil Steril;103(2):455-61.	2015	4,42	

Neoplasia, general	Ghaedi H, Bastami M, Zare-Abdollahi D, Alipoor B, Movafagh A, Mirfakhraie R, Omrani MD, Masotti A	Bioinformatics prioritization of SNPs perturbing microRNA regulation of hematological malignancy-implicated genes	Genomics. 106(6):360-6.	2015	2,28	
Neoplasia, general	Giordano P, Saracco P, Grassi M, Luciani M, Banov L, Carraro F, Crocoli A, Cesaro S, Zanazzo GA, Molinari AC; Italian Association of Pediatric Hematology and Oncology (AIEOP)	Recommendations for the use of long-term central venous catheter (CVC) in children with hemato-oncological disorders: management of CVC-related occlusion and CVC-related thrombosis. On behalf of the coagulation defects working group and the supportive therapy working group of the Italian Association of Pediatric Hematology and Oncology (AIEOP)	Ann Hematol. 94(11):1765-76.	2015	2,63	AIEOP
Neoplasia, general	Crocoli A, Martucci C, Leopardi E, Padua M, Serra A, Cacchione A, Coletti V, Palumbo G, Ciofi degli Atti ML, Ravà L, Inserra A	A dedicated protocol and environment for central venous catheter removal in pediatric patients affected by onco-hematological diseases	J Vasc Access. 15(6):486-91.	2014	1,02	
Neoplasia, general	Crocoli A, Tornesello A, Pittiruti M, Barone A, Muggeo P, Inserra A, Molinari AC, Grillenzoni V, Durante V, Cicalese MP, Zanazzo GA, Cesaro S.	Central venous access devices in pediatric malignancies: a position paper of Italian Association of Pediatric Hematology and Oncology.	J Vasc Access; 16(2):130-6.	2015	1,53	AIEOP
Neoplasia, general	Ferrari A, Rondelli R, Pession A, Mascarin M, Buzzoni C, Mosso ML, Maule M, Barisone E, Bertolotti M, Clerici CA, Jankovic M, Fagioli F, Biondi A	Adolescents with Cancer in Italy: Improving Access to National Cooperative Pediatric Oncology Group (AIEOP) Centers	Pediatr Blood Cancer; 63(6):1116-9.	2016	2,63	AIEOP
Neoplasia, general	Rondelli R, Jankovic M, Soresina A, Valsecchi MG, De Rosa M, Cuttini M, Haupt R, Aricò M, Bisogno G, Locatelli F, Magnani C, Merletti F, Zecca M, Pession A	The contribution of the Italian Association of paediatric haematology and oncology (AIEOP)	Epidemiol Prev. Sep-Oct;40(5Suppl2):23-27	2016	0,90	AIEOP

Neoplasia, general	Caselli D, Petris MG, Rondelli R, Carraro F, Colombini A, Muggeo P, Ziino O, Melchionda F, Russo G, Pierani P, Soncini E, DeSantis R, Zanazzo G, Barone A, Cesaro S, Cellini M, Mura R, Milano GM, Meazza C, Cicalese MP, Tropia S, De Masi S, Castagnola E, Aricò M; Infectious Diseases Working Group of the Associazione Italiana Ematologia Oncologia Pediatrica.	Single-day trimethoprim/sulfamethoxazole prophylaxis for Pneumocystis pneumonia in children with cancer	J Pediatr; 164(2):389-92	2014	3,89	AIEOP
Neoplasia, general	Spreafico F, Bongarzone I, Pizzamiglio S, Magni R, Taverna E, De Bortoli M, Ciniselli CM, Barzanò E, Biassoni V, Luchini A, Liotta LA, Zhou W, Signore M, Verderio P, Massimino M	Proteomic analysis of cerebrospinal fluid from children with central nervous system tumors identifies candidate proteins relating to tumor metastatic spread	Oncotarget. doi: 10.18632/oncotarget.17579	2017	5,00	
Neoplasia, general	Indolfi P, Picazio S, Perrotta S, Rossi F, Pession A, Di Martino M, Pota E, Di Pinto D, Indolfi C, Rondelli R, Vetrano F, Casale F.	Time trends of cancer incidence in childhood in Campania region: 25 years of observation	Ital J Pediatr. Sep 6;42(1):82.	2016	1,61	
Neoplasia, general	D'Ambrosio G, Del Prete L, Grimaldi C, Bertocchini A, Lo Zupone C, Monti L, De Ville De Goyet J	Pancreaticoduodenectomy for malignancies in children	J Pediatr Surg. 49(4):534-8.	2014	1,31	
Neoplasia, general	De Ioris MA, Carai A, Valente P, Angioni A, Randisi F, Cozza R, Romanzo A, Marras CE, Mastronuzzi A	Sporadic Retinoblastoma and Pilocytic Astrocytoma: A Rare Association of Two Tumors	Pediatr Blood Cancer. 62(12):2245-6.	2015	2,39	
Neoplasia, general	Caselli D, Cesaro S, Fagioli F, Carraro F, Ziino O, Zanazzo G, Meazza C, Colombini A, Castagnola E; Infectious Diseases Study Group of the Italian Association of Pediatric Hematology and Oncology (AIEOP).	Incidence of colonization and bloodstream infection with carbapenem-resistant Enterobacteriaceae in children receiving antineoplastic chemotherapy in Italy.	Infect Dis (Lond); 48(2):152-5.	2016	1,36	AIEOP

Neoplasia, general	Fruci D, Romania P, D'Alicandro V, Locatelli F	Endoplasmic reticulum aminopeptidase 1 function and its pathogenic role in regulating innate and adaptive immunity in cancer and major histocompatibility complex class I-associated autoimmune diseases	Tissue Antigens. 84(2):177-86.	2014	2,35	
Neoplasia, general	Bizzarri C, Pinto R, Ciccone S, Brescia LP, Locatelli F, Cappa M	Early and progressive insulin resistance in young, non-obese cancer survivors treated with hematopoietic stem cell transplantation	Pediatr Blood Cancer. 62(9):1650-5.	2015	2,39	
Neoplasia, general	Muccio L, Bertaina A, Falco M, Pende D, Meazza R, Lopez-Botet M, Moretta L, Locatelli F, Moretta A, Della Chiesa M	Analysis of memory-like Natural Killer cells in Human Cytomegalovirus-infected children undergoing alphabeta+T- and B-cell depleted HSCT for hematological malignancies	Haematologica. 2016 Mar;101(3):371-81.	2015	5,81	
Neoplasia, general	Cusinato M, Calvo V, Bisogno G, Viscardi E, Pillon M, Opocher E, Basso G, Montanaro M.	Attachment orientations and psychological adjustment of parents of children with cancer: A matched-group comparison.	J Psychosoc Oncol. doi: 10.1080/07347332.2017.1335365.	2017	1,29	
Neoplasia, general	Proserpio T, Veneroni L, Silva M, Lassaletta A, Lorenzo R, Magni C, Bertolotti M, Barisone E, Mascarin M, Jankovic M, D'Angelo P, Clerici CA, Garrido-Colino C, Gutierrez-Carrasco I, Echebarria A, Biondi A, Massimino M, Casale F, Tamburini A, Ferrari A.	Spiritual support for adolescent cancer patients: a survey of pediatric oncology centers in Italy and Spain.	Tumori. 102(4):376-80.	2016	1,07	
Neoplasia, general	Castagnola E, Bagnasco F, Amoroso L, Caviglia I, Caruso S, Faraci M, Calvillo M, Moroni C, Bandettini R, Cangemi G, Magnano GM, Buffa P, Moscatelli A, Haupt R	Role of management strategies in reducing mortality from invasive fungal disease in children with cancer or receiving hemopoietic stem cell transplant: a single center 30-year experience.	Pediatr Infect Dis J. 33(3):233-7.	2014	5,72	

Neoplasia, general	Castagnola E, Dallorso S, Haupt R.	Administration schedule and ototoxicity of amikacin in children with cancer.	Pediatr Blood Cancer. 61(2):192.	2014	2,63	
Neoplasia, general	Terenziani M, Spinelli M, Jankovic M, Bardi E, Hjorth L, Haupt R, Michel G, Byrne J; PanCare Network.	Practices of pediatric oncology and hematology providers regarding fertility issues: a European survey.	Pediatr Blood Cancer. 61(11):2054-8.	2014	2,63	
Neoplasia, general	Stark D, Bielack S, Brugieres L, Dirksen U, Duarte X, Dunn S, Erdelyi DJ, Grew T, Hjorth L, Jazbec J, Kabickova E, Konsoulova A, Kowalczyk JR, Lassaletta A, Laurence V, Lewis I, Monrabal A, Morgan S, Mountzios G, Olsen PR, Renard M, Saeter G, van der Graaf WT, Ferrari A	Teenagers and young adults with cancer in Europe: from national programmes to a European integrated coordinated project	Eur J Cancer Care; 25(3):419-27.	2016	1,79	
Neoplasia, general	van Dorp W, Mulder RL, Kremer LC, Hudson MM, van den Heuvel-Eibrink MM, van den Berg MH, Levine JM, van Dulmen-den Broeder E, di Iorgi N, Albanese A, Armenian SH, Bhatia S, Constine LS, Corrias A, Deans R, Dirksen U, Gracia CR, Hjorth L, Kroon L, Lambalk CB, Landier W, Levitt G, Leiper A, Meacham L, Mussa A, Neggers SJ, Oeffinger KC, Revelli A, van Santen HM, Skinner R, Toogood A, Wallace WH, Haupt R.	Recommendations for Premature Ovarian Insufficiency Surveillance for Female Survivors of Childhood, Adolescent, and Young Adult Cancer: A Report From the International Late Effects of Childhood Cancer Guideline Harmonization Group in Collaboration With the PanCareSurFup Consortium.	J Clin Oncol. 34(28):3440-50.	2016	20,98	
Neoplasia, general	Castagnola E, Caviglia I, Pescetto L, Bagnasco F, Haupt R, Bandettini R.	Antibiotic susceptibility of Gram-negatives isolated from bacteremia in children with cancer. Implications for empirical therapy of febrile neutropenia.	Future Microbiol. 10(3):357-64.	2015	3,64	

Neoplasia, general	Tremolada M, Bonichini S, Basso G, Pillon M.	Post-traumatic Stress Symptoms and Post-traumatic Growth in 223 Childhood Cancer Survivors: Predictive Risk Factors.	Front Psychol. 7:287.	2016	3,32	
Neoplasia, general	Magnani C, Mattioli S, Miligi L, Ranucci A, Rondelli R, Salvan A, Bisanti L, Masera G, Rizzari C, Zambon P, Cannizzaro S, Gafà L, Luzzatto LL, Benvenuti A, Michelozzi P, Kirchmayer U, Cocco P, Biddau P, Galassi C, Celentano E, Guarino E, Assennato G, de Nichilo G, Merlo DF, Bocchini V, Pannelli F, Mosciatti P, Minelli L, Chiavarini M, Cuttini M, Casotto V, Torregrossa MV, Valenti RM, Forastiere F, Haupt R, Lagorio S, Risica S, Polichetti A.	SETIL: Italian multicentric epidemiological case-control study on risk factors for childhood leukaemia, non hodgkin lymphoma and neuroblastoma: study population and prevalence of risk factors in Italy.	Ital J Pediatr. 40:103.	2014	1,61	
Neoplasia, general	Faraci M, Bagnasco F, Giardino S, Conte M, Micalizzi C, Castagnola E, Lampugnani E, Moscatelli A, Franceschi A, Carcillo JA, Haupt R.	Intensive care unit admission in children with malignant or nonmalignant disease: incidence, outcome, and prognostic factors: a single-center experience.	J Pediatr Hematol Oncol. 36(7):e403-9.	2014	1,15	
Neoplasia, general	Bagattoni S, D'Alessandro G, Prete A, Piana G, Pession A.	Oral health and dental late adverse effects in children in remission from malignant disease. A pilot case-control study in Italian children.	Eur J Paediatr Dent. 15(1):45-50.	2014	0,42	
Neoplasia, general	Tremolada M, Bonichini S, Basso G, Pillon M.	Perceived social support and health-related quality of life in AYA cancer survivors and controls.	Psychooncology. 25(12):1408-1417.	2016	3,26	
Neoplasia, general	Cordelli DM, Masetti R, Zama D, Toni F, Castelli I, Ricci E, Franzoni E, Pession A.	Central Nervous System Complications in Children Receiving Chemotherapy or Hematopoietic Stem Cell Transplantation.	Front Pediatr. 5:105.	2017	2,17	

Neoplasia, general	Pagano E, Baldi I, Mosso ML, di Montezemolo LC, Fagioli F, Pastore G, Merletti F.	The economic burden of caregiving on families of children and adolescents with cancer: a population-based assessment.	Pediatr Blood Cancer. 61(6):1088-93.	2014	2,63	
Neoplasia, general	Felicetti F, D'Ascenzo F, Moretti C, Corrias A, Omedè P, Marra WG, Arvat E, Fagioli F, Brignardello E, Gaita F.	Prevalence of cardiovascular risk factors in long-term survivors of childhood cancer: 16 years follow up from a prospective registry.	Eur J Prev Cardiol. 22(6):762-70.	2015	3,36	
Neoplasia, general	Scarponi D, Pession A.	Play Therapy to Control Pain and Suffering in Pediatric Oncology.	Front Pediatr. 4:132.	2016	2,17	
Neoplasia, general	Brignardello E, Felicetti F, Castiglione A, Fortunati N, Matarazzo P, Biasin E, Sacerdote C, Ricardi U, Fagioli F, Corrias A, Arvat E.	GH replacement therapy and second neoplasms in adult survivors of childhood cancer: a retrospective study from a single institution.	J Endocrinol Invest. 38(2):171-6.	2015	1,99	
Neoplasia, general	Zucchetti G, Bellini S, Bertolotti M, Bona F, Biasin E, Bertorello N, Tirtei E, Fagioli F.	Body Image Discomfort of Adolescent and Young Adult Hematologic Cancer Survivors.	J Adolesc Young Adult Oncol.Jun;6(2):377-380.	2017	0,05	
Neoplasia, general	Zucchetti G, Bertorello N, Angelastro A, Gianino P, Bona G, Barbara A, Besenzon L, Brach Del Prever A, Pesce F, Nangeroni M, Fagioli F.	Improving healthcare in pediatric oncology: development and testing of multiple indicators to evaluate a hub-and-spoke model.	Tumori. Jun 6:0. doi: 10.5301/tj.5000645.	2017	1,07	
Neoplasia, general	Brignardello E, Felicetti F, Castiglione A, Nervo A, Biasin E, Ciccone G, Fagioli F, Corrias A.	Gonadal status in long-term male survivors of childhood cancer.	J Cancer Res Clin Oncol. 142(5):1127-32.	2016	3,14	
Neoplasia, general	Brignardello E, Felicetti F, Castiglione A, Gallo M, Maletta F, Isolato G, Biasin E, Fagioli F, Corrias A, Palestini N.	Ultrasound surveillance for radiation-induced thyroid carcinoma in adult survivors of childhood cancer.	Eur J Cancer. 55:74-80.	2016	6,16	
Neoplasia, general	Pietra G, Vitale C, Pende D, Bertaina A, Moretta F, Falco M, Vacca P, Montaldo E, Cantoni C, Mingari MC, Moretta A, Locatelli F, Moretta L	Human natural killer cells: news in the therapy of solid tumors and high-risk leukemias	Cancer Immunol Immunother. 65(4):465-76.	2016	3,94	

Neoplasia, general	Bizzarri C, Pinto R, Ciccone S, Brescia LP, Locatelli F, Cappa M	Early and progressive insulin resistance in young, non-obese cancer survivors treated with hematopoietic stem cell transplantation	Pediatr Blood Cancer. 62(9):1650-5.	2015	2,39	
Neoplasia, general	Muccio L, Bertaina A, Falco M, Pende D, Meazza R, Lopez-Botet M, Moretta L, Locatelli F, Moretta A, Della Chiesa M	Analysis of memory-like Natural Killer cells in Human Cytomegalovirus-infected children undergoing alphabeta+T- and B-cell depleted HSCT for hematological malignancies	Haematologica. 2016 Mar;101(3):371-81.	2015	5,81	
Neoplasia, general	Potenza L, Vallerini D, Barozzi P, Riva G, Gilioli A, Forghieri F, Candoni A, Cesaro S, Quadrelli C, Maertens J, Rossi G, Morselli M, Codeluppi M, Mussini C, Colaci E, Messerotti A, Paolini A, Maccaferri M, Fantuzzi V, Del Giovane C, Stefani A, Morandi U, Maffei R, Marasca R, Narni F, Fanin R, Comoli P, Romani L, Beauvais A, Viale PL, Latgè JP, Lewis RE, Luppi M	Mucorales-Specific T Cells in Patients with Hematologic Malignancies	PLoS One. 2016 Feb 12;11(2):e0149108	2016	4,41	
Neoplasia, general	Ferrari A	SIAMO: Italian Pediatric Oncologists and Adult Medical Oncologists Join Forces for Adolescents with Cancer	Pediatr Hematol Oncol. 2014;31(6):574-5.	2014	1,09	
Neoplasia, general	Stabile H, Nisti P, Morrone S, Pagliara D, Bertaina A, Locatelli F, Santoni A, Gismondi A	Multifunctional human CD56 low CD16 low natural killer cells are the prominent subset in bone marrow of both healthy pediatric donors and leukemic patients	Haematologica. 100(4):489-98.	2015	5,81	
Neuroblastic tumors	Avanzini S, Faticato MG, Crocoli A, Virgone C, Viglio C, Severi E, Fagnani AM, Cecchetto G, Riccipetitoni G, Noccioli B, Leva E, Sementa A R,Mattioli G, Inserra A	Comparative retrospective study on the modalities of biopsying peripheral neuroblastic tumors: a report from the Italian Pediatric Surgical Oncology Group (GICOP)	Pediatr Blood Cancer. 64(5).	2017	2,63	

Neuroblastic tumors	Calabrese FM, Clima R, Pignataro P, Lasorsa VA, Hogarty MD, Castellano A, Conte M, Tonini GP, Iolascon A, Gasparre G, Capasso M	A comprehensive characterization of rare mitochondrial DNA variants in neuroblastoma	Oncotarget. 7(31):49246-49258.	2016	5,01	
Neuroblastic tumors	Amoroso L, Erminio G, Makin G, Pearson AD, Brock P, Valteau-Couanet D, Castel V, Pasquet M, Laureys G, Thomas C, Luksch R, Ladenstein R, Haupt R, Garaventa A; SIOPEN Group	Topotecan-Vincristine-Doxorubicin in Stage 4 High Risk Neuroblastoma Patients Failing to Achieve a Complete Metastatic Response to Rapid COJEC - a SIOPEN Study	Cancer Res Treat. Mar 21. doi: 10.4143/crt.2016.511	2017	4,24	
Neuroblastic tumors	De Bernardi B, Quaglietta L, Haupt R, Castellano Aurora, Tirtei E, Luksch R, Mastrangelo S, Viscardi E, Indolfi P, Cellini M, Tamburini A, Erminio G, Gandolfo C, Sorrentino S, Vetrella S, Gigliotti A.	Neuroblastoma with symptomatic epidural compression in the infant: The AIEOP experience	Pediatr Blood Cancer. 61(8):1369-75.	2014	2,56	AIEOP
Neuroblastic tumors	Avanzini S, Pio L, Erminio G, Granata C, Holmes K, Gambart M, Buffa P, Castel V, Valteau Couanet D, Garaventa A, Pistorio A, Cecchetto G, Martucciello G, Mattioli G, Sarnacki S	Image-defined risk factors in unresectable neuroblastoma: SIOPEN study on incidence, chemotherapy-induced variation, and impact on surgical outcomes	Pediatr Blood Cancer. 2017 Apr 25. doi: 10.1002/pbc.26605	2017	2,63	
Neuroblastic tumors	De Ioris MA, Crocoli A, Contoli B, Garganese MC, Natali G, Tomà P, Jenkner A, Boldrini R, De Pasquale MD, Milano GM, Madafferi S, Castellano A, Locatelli F, Inserra A	Local control in metastatic neuroblastoma in children over 1 year of age	BMC Cancer. 15:79.	2015	3,36	
Neuroblastic tumors	Di Cataldo A, Agodi A, Balaguer J, Garaventa A, Barchitta M, Segura V, Bianchi M, Castel V, Castellano A, Cesaro S, Couselo JM, Cruz O, D'Angelo P, De Bernardi B, Donat J, de Andoin NG, Hernandez MI, La Spina M, Lillo M, Lopez-Almaraz R, Luksch R, Mastrangelo S, Mateos E, Molina J, Moscheo C, Mura R, Porta F, Russo G, Tondo A, Torrent M, Vetrella S, Villegas JA, Viscardi E, Zanazzo GA, Cañete A	Metastatic neuroblastoma in infants: are survival rates excellent only within the stringent framework of clinical trials?	Clin Transl Oncol. 19(1):76-83.	2017	2,08	

Neuroblastic tumors	Defferrari R, Mazzocco K, Ambros IM, Ambros PF, Bedwell C, Beiske K, Bénard J, Berbegall AP, Bown N, Combaret V, Couturier J, Erminio G, Gambini C, Garaventa A, Gross N, Haupt R, Kohler J, Jeison M, Lunec J, Marques B, Martinsson T, Noguera R, Parodi S, Schleiermacher G, Tweddle DA, Valent A, Van Roy N, Vicha A, Villamon E, Tonini GP	Influence of segmental chromosome abnormalities on survival in children over the age of 12 months with unresectable localized peripheral neuroblastic tumours without MYCN amplification	Br J Cancer;112(2):290-5	2015	5,56	
Neuroblastic tumors	Dondero A, Pastorino F, Della Chiesa M, Corrias MV, Morandi F, Pistoia V, Olive D, Bellora F, Locatelli F, Castellano A, Moretta L, Moretta A, Bottino C, Castriconi R	PD-L1 expression in metastatic neuroblastoma as an additional mechanism for limiting immune surveillance	Oncoimmunology. 5(1):e1064578.	2015	7,64	
Neuroblastic tumors	Furfaro AL, Piras S, Domenicotti C, Fenoglio D, De Luigi A, Salmona M, Moretta L, Marinari UM, Pronzato MA, Traverso N, Nitti M	Role of Nrf2, HO-1 and GSH in Neuroblastoma Cell Resistance to Bortezomib	PLoS One. 11(3):e0152465	2016	3,06	
Neuroblastic tumors	Gigliotti AR, De Ioris MA, De Grandis E, Podda M, Cellini M, Sorrentino S, De Bernardi B, Paladini D, Gandolfo C	Congenital neuroblastoma with symptoms of epidural compression at birth	Pediatr Hematol Oncol. 33(2):94-101.	2016	1,09	
Neuroblastic tumors	Mazzocco K, Defferrari R, Sementa AR, Garaventa A, Longo L, De Mariano M, Esposito MR, Negri F, Ircolò D, Viscardi E, Luksch R, D'Angelo P, Prete A, Castellano A, Massirio P, Erminio G, Gigliotti AR, Tonini GP, Conte M	Genetic abnormalities in adolescents and young adults with neuroblastoma: A report from the Italian Neuroblastoma group	Pediatr Blood Cancer. 62(10):1725-32.	2015	2,39	
Neuroblastic tumors	Marino S, La Spina M, Scuderi MG, Di Benedetto V, Magro G, Belfiore G, Coronella M, D'Amico S, Lo Nigro L, Russo G, Di Cataldo A	Bilateral adrenal neuroblastoma in the infant: Is it an image-defined risk factor?	Pediatr Hematol Oncol. May;33(4):259-63	2016	1,09	
Neuroblastic tumors	Mina M, Boldrini R, Citti A, Romania P, D'Alicandro V, De Ioris M, Castellano A, Furlanello C, Locatelli F, Fruci D	Tumor-infiltrating T lymphocytes improve clinical outcome of therapy-resistant neuroblastoma	Oncoimmunology. 4(9):e1019981.	2015	6,27	

Neuroblastic tumors	Zama D, Morello W, Masetti R, Cordelli DM, Massaccesi E, Prete A, Pession A.	Inflammatory disease of the central nervous system induced by anti-GD2 monoclonal antibody in a patient with high risk neuroblastoma.	Pediatr Blood Cancer. 61(8):1521-2.	2014	2,63	
Neuroblastic tumors	Parodi S, Merlo DF, Ranucci A, Miligi L, Benvenuti A, Rondelli R, Magnani C, Haupt R; SETIL Working Group.	Risk of neuroblastoma, maternal characteristics and perinatal exposures: the SETIL study.	Cancer Epidemiol. 38(6):686-94.	2014	2,64	
Neuroblastic tumors	Vallero SG, Lijoi S, Bertin D, Pittana LS, Bellini S, Rossi F, Peretta P, Basso ME, Fagioli F.	End-of-life care in pediatric neuro-oncology.	Pediatr Blood Cancer. 61(11):2004-11.	2014	2,63	
Neuroblastic tumors	Amoroso L, Erminio G, Makin G, Pearson AD, Brock P, Valteau-Couanet D, Castel V, Pasquet M, Laureys G, Thomas C, Luksch R, Ladenstein R, Haupt R, Garaventa A; SIOPEN Group.	Topotecan-Vincristine-Doxorubicin in Stage 4 High Risk Neuroblastoma Patients Failing to Achieve a Complete Metastatic Response to Rapid COJEC - a SIOPEN Study.	Cancer Res Treat. doi: 10.4143/crt.2016.511.	2017	4,43	
Neuroblastic tumors	Stigliani S, Scaruffi P, Lagazio C, Persico L, Carlini B, Varesio L, Morandi F, Morini M, Gigliotti AR, Esposito MR, Viscardi E, Cecinati V, Conte M, Corrias MV	Deregulation of focal adhesion pathway mediated by miR-659-3p is implicated in bone marrow infiltration of stage M neuroblastoma patients	Oncotarget. 6(15):13295-308	2015	5,01	
Neuroblastic tumors	Pezzolo A, Pistorio A, Gambini C, Haupt R, Ferraro M, Erminio G, De Bernardi B, Garaventa A, Pistoia V.	Intratumoral diversity of telomere length in individual neuroblastoma tumors.	Oncotarget. 6(10):7493-503.	2015	5,01	
Neuroblastic tumors	Defferrari R, Mazzocco K, Ambros IM, Ambros PF, Bedwell C, Beiske K, Bénard J, Berbegall AP, Bown N, Combaret V, Couturier J, Erminio G, Gambini C, Garaventa A, Gross N, Haupt R, Kohler J, Jeison M, Lunec J, Marques B, Martinsson T, Noguera R, Parodi S, Schleiermacher G, Tweddle DA, Valent A, Van Roy N, Vicha A, Villamon E, Tonini GP.	Influence of segmental chromosome abnormalities on survival in children over the age of 12 months with unresectable localised peripheral neuroblastic tumours without MYCN amplification.	Br J Cancer. 112(2):290-5.	2015	5,57	

Neuroblastic tumors	De Bernardi B, Quaglietta L, Haupt R, Castellano A, Tirtei E, Luksch R, Mastrangelo S, Viscardi E, Indolfi P, Cellini M, Tamburini A, Erminio G, Gandolfo C, Sorrentino S, Vetrella S, Gigliotti AR.	Neuroblastoma with symptomatic epidural compression in the infant: the AIEOP experience.	Pediatr Blood Cancer. 61(8):1369-75.	2014	2,63	
Neuroblastic tumors	Morandi F, Pozzi S, Barco S, Cangemi G, Amoroso L, Carlini B, Pistoia V, Corrias MV	CD4+CD25hiCD127- Treg and CD4+CD45R0+CD49b+LAG3+ Tr1 cells in bone marrow and peripheral blood samples from children with neuroblastoma	Oncoimmunology. 5(12):e1249553.	2016	7,64	
Neuroblastic tumors	Avanzini S, Faticato MG, Sementa AR, Granata C, Martucciello G, Pio L, Prato AP, Garaventa A, Bisio G, Montobbio G, Buffa P, Mattioli G	Video-Assisted Needle Core Biopsy in Children Affected by Neuroblastoma: A Novel Combined Technique	Eur J Pediatr Surg; 27(2):166-170	2017	1,26	
Neuroblastic tumors	Formicola D, Petrosino G, Lasorsa VA, Pignataro P, Cimmino F, Vetrella S, Longo L, Tonini GP, Oberthuer A, Iolascon A, Fischer M, Capasso M	An 18 gene expression-based score classifier predicts the clinical outcome in stage 4 neuroblastoma	J Transl Med; 14(1):142	2016	3,69	
Neuroblastic tumors	Veschi V, Petroni M, Bartolazzi A, Altavista P, Dominici C, Capalbo C, Boldrini R, Castellano A, McDowell H, Pizer B, Frati L, Screpanti I, Gulino A, Giannini G	Galectin-3 is a marker of favorable prognosis and a biologically relevant molecule in neuroblastic tumors	Cell Death Dis. 5:e1100.	2014	5,18	
Neurocytoma	Messina R, Cefalo M, Cappelletti S, Rebessi E, Carai A, Colafati G, Diomedi Camassei F, Cacchione A, Marras CE, Mastronuzzi A	Behavioral disorders as unusual presentation of pediatric extraventricular neurocytoma: report on two cases and review of the literature	BMC Neurol. 14:242.	2014	2,49	
Osteosarcoma	Tirtei E, Asaftei SD, Manicone R, Cesari M, Paioli A, Rocca M, Ferrari S, Fagioli F.	Survival after second and subsequent recurrences in osteosarcoma: a retrospective multicenter analysis.	Tumori. May 10:0. doi: 10.5301/tj.5000636.	2017	1,07	

Osteosarcoma	Ferrari S, Meazza C, Palmerini E, Tamburini A, Fagioli F, Cozza R, Ferraresi V, Bisogno G, Mascarin M, Cefalo G, Manfrini M, Capanna R, Biagini R, Donati D, Picci P.	Nonmetastatic osteosarcoma of the extremity. Neoadjuvant chemotherapy with methotrexate, cisplatin, doxorubicin and ifosfamide. An Italian Sarcoma Group study (ISG/OS-Oss).	Tumori. 100(6):612-9.	2014	1,07	
Osteosarcoma	Palmerini E, Jones RL, Marchesi E, Paioli A, Cesari M, Longhi A, Meazza C, Coccoli L, Fagioli F, Asaftei S, Grignani G, Tamburini A, Pollack SM, Picci P, Ferrari S.	Gemcitabine and docetaxel in relapsed and unresectable high-grade osteosarcoma and spindle cell sarcoma of bone.	BMC Cancer. 16:280.	2016	3,27	
Osteosarcoma	Grignani G, Palmerini E, Ferraresi V, D'Ambrosio L, Bertulli R, Asaftei SD, Tamburini A, Pignochino Y, Sangiolo D, Marchesi E, Capozzi F, Biagini R, Gambarotti M, Fagioli F, Casali PG, Picci P, Ferrari S, Aglietta M; Italian Sarcoma Group.	Sorafenib and everolimus for patients with unresectable high-grade osteosarcoma progressing after standard treatment: a non-randomised phase 2 clinical trial.	Lancet Oncol; 16(1):98-107.	2015	26,50	
Osteosarcoma	Ferrari S, Meazza C, Palmerini E, Cozza R, Biagini R, Donati D, Picci P	Nonmetastatic osteosarcoma of the extremity. Neoadjuvant chemotherapy with methotrexate, cisplatin, doxorubicin and ifosfamide. An Italian Sarcoma Group study (ISG/OS-Oss)	Tumori. 100(6):612-9.	2014	1,09	
Ewing sarcoma	Paioli A, Luksch R, Fagioli F, Tamburini A, Cesari M, Palmerini E, Abate ME, Marchesi E, Balladelli A, Pratelli L, Ferrari S.	Chemotherapy-related toxicity in patients with non-metastatic Ewing sarcoma: influence of sex and age.	J Chemother. 26(1):49-56.	2014	1,33	
Ewing sarcoma	Ferrari S, Luksch R, Hall KS, Fagioli F, Prete A, Tamburini A, Tienghi A, DiGirolamo S, Paioli A, Abate ME, Podda M, Cammelli S, Eriksson M, Brach del Prever A.	Post-relapse survival in patients with Ewing sarcoma.	Pediatr Blood Cancer. 62(6):994-9.	2015	2,63	

other	De Pasquale MD, Crocoli A, Conte M, Indolfi P, D'Angelo P, Boldrini R, Terenziani M, Inserra A	Mediastinal Germ Cell Tumors in Pediatric Patients: A Report From the Italian Association of Pediatric Hematology and Oncology	Pediatr Blood Cancer. 63(5):808-12.	2016	2,63	AIEOP
other	Terenziani M, Bisogno G, Boldrini R, Cecchetto G, Conte M, Boschetti L, De Pasquale MD, Biasoni D, Inserra A, Siracusa F, Basso ME, De Leonardis F, Di Pinto D, Barretta F, Spreafico F, D'Angelo P.	Malignant ovarian germ cell tumors in pediatric patients: The AIEOP (Associazione Italiana Ematologia Oncologia Pediatrica) study.	Pediatr Blood Cancer. 2017 Apr 27. doi: 10.1002/pbc.26568	2017	2,63	AIEOP
other	Bizzarri C and Bottaro G	Endocrine implications of neurofibromatosis 1 in childhood	Horm Res Paediatr. 83(4):232-41.	2015	1,57	
other	Diociaiuti A, Guidi B, Surrenti T, Boldrini R, Callea F, El Hachem M	A rare case of segmental neurofibromatosis with multiple blue-red pseudoatrophic plaques	Cutis. 94(3):149-52.	2014	0,59	
other	Giovannoni I, Callea F, Boldrini R, Inserra A, Cozza R, Francalanci P	Malignant pheochromocytoma in a 16-year-old patient with neurofibromatosis type 1	Pediatr Dev Pathol. 17(2):126-9.	2014	0,86	
other	Pinna V, Lanari V, Daniele P, Digilio MC, Dallapiccola B, Clementi M, Tartaglia M, De Luca A	p.Arg1809Cys substitution in neurofibromin is associated with a distinctive NF1 phenotype without neurofibromas	Eur J Hum Genet. 23(8):1068-71.	2015	4,23	
other	Pizzi M, di Lorenzo I, d'Amore ES, D'Angelo P, Alaggio R.	Pediatric gastrointestinal PEComas: a diagnostic challenge.	Pediatr Dev Pathol. 17(5):406-8.	2014	0,77	
other	Astolfi A, Melchionda F, Perotti D, Fois M, Indio V, Urbini M, Genovese CG, Collini P, Salfi N, Nantron M, D'Angelo P, Spreafico F, Pession A.	Whole transcriptome sequencing identifies BCOR internal tandem duplication as a common feature of clear cell sarcoma of the kidney.	Oncotarget. 6(38):40934-9.	2015	5,01	
other	De Pasquale MD, Mastronuzzi A, De Sio L, Serra A, Grimaldi C, Chinali M, Giordano U	Transient global ventricular dysfunction in an adolescent affected by pancreatic adenocarcinoma	BMC Pediatr. 16:99.	2016	1,81	

other	Maggioli C, Cambiaso P, Ciccone S, Colafati GS, Cappa M	Long-term first line medical treatment in a 4-year-old girl with Xq26.3 microduplication-negative somatotropinoma. Case report and literature review	J Pediatr Endocrinol Metab. 29(4):497-501.	2016	0,91	
other	Massi D, Tomasini C, Senetta R, Boldrini R, Gambini C, Montesco MC	Atypical Spitz tumors in patients younger than 18 years	J Am Acad Dermatol. 72(1):37-46.	2015	5,00	
other	Chiaravalli S, Guzzo M, Bisogno G, De Pasquale MD, Casanova M, Cecchetto G, Ferrari A	Salivary gland carcinomas in children and adolescents: The Italian TREP project experience	Pediatr Blood Cancer. 61(11):1961-8.	2014	2,56	
other	Specchio N, Rizzi M, Fusco L, Rebessi E, Cappelletti S, De Palma L, Savioli A, De Benedictis A, Marras CE, Vigevano F, Delalande O	Acute intralesional recording in hypothalamic hamartoma: description of 4 cases	Acta Neurol Belg. 115(3):233-9.	2015	0,60	
other	Morbini P, Capello GL, Alberizzi P, Benazzo M, Paglino C, Comoli P, Pedrazzoli P	Markers of squamocolumnar junction cells in normal tonsils and oropharyngeal cancer with and without HPV infection	Histol Histopathol. Jul;30(7):833-9	2015	1,87	
other	Cetrano E, Polito A, Carotti A	Primitive intrapericardial teratoma associated with yolk sac tumour	Cardiol Young. 25(1):158-60.	2015	0,86	
other	Angelino G, De Pasquale MD, De Sio L, Serra A, Massimi L, De Vito R, Marrazzo A, Lancella L, Carai A, Antonelli M, Giangaspero F, Gessi M, Menchini L, Scarciolla L, Longo D, Mastronuzzi A	NRAS(Q61K) mutated primary leptomeningeal melanoma in a child: case presentation and discussion on clinical and diagnostic implications	BMC Cancer. 16:512.	2016	3,27	
other	Ferrari A, Bisogno G, Cecchetto G, Santinami M, Maurichi A, Bono A, Vajna De Pava M, Pierani P, Bertolini P, Rossi CR, De Salvo GL	Cutaneous melanoma in children and adolescents: the Italian rare tumors in pediatric age project experience	J Pediatr. 2014 Feb;164(2):376-82	2014	0,52	
other	Rapini N, Lidano R, Pietrosanti S, Vitiello G, Grimaldi Chiara, Postorivo D, Nardone A, Del Bufalo F, Brancati F, Manca Bitti M.	De Novo 13q13.3-21.31 deletion involving RB1 gene in a patient with hemangioendothelioma of the liver	Ital J Pediatr. 40:5.	2014	1,24	

other	Indini A, Bisogno G, Cecchetto G, Vitellaro M, Signoroni S, Massimino M, Riccipetitoni G, Zecca M, Dall'Igna P, Debora De Pasquale M, Inserra A, Chiaravalli S, Basso E, Virgone C, Sorbara S, Di Bartolomeo M, D'Angelo P, Ferrari A.	Gastrointestinal tract carcinoma in pediatric and adolescent age: The Italian TREP project experience.	Pediatr Blood Cancer doi:10.1002/pbc.26658	2017	2,63	
other	Nobili V, Alisi A, Grimaldi C, Liccardo D, Francalanci P, Monti L, Castellano A, De Ville De Goyet J	Non-alcoholic fatty liver disease and hepatocellular carcinoma in a 7-year-old obese boy: Coincidence or comorbidity?	Pediatr Obes. 9(5):e99-e102.	2014	2,42	
other	Carrega P, Loiacono F, Di Carlo E, Moretta L, Ferlazzo G	NCR(+)ILC3 concentrate in human lung cancer and associate with intratumoral lymphoid structures	Nat Commun. 6:8280.	2015	11,47	
other	Cefalo M, Colafati G, Romanzo A, Modugno A, De Vito R, Mastronuzzi A	Congenital cystic eye associated with a low-grade cerebellar lesion that spontaneously regressed	BMC Ophthalmol. 14:80.	2014	1,08	
other	Antonelli M, Korshunov A, Mastronuzzi A, Diomedi Camassei F, Carai A, Giangaspero F	Long-term survival in a case of ETANTR with histological features of neuronal maturation after therapy	Virchows Arch. 466(5):603-7.	2015	2,65	
other	Diociaiuti A, Inserra A, Fuertes De Vega I, Rota C, Surrenti T, Giraldi R, Piemontese MR, Giovannoni I, Callea F, El Hachem M	Naevoid Basal Cell Carcinoma Syndrome in a 22-month-old Child Presenting with Multiple Basal Cell Carcinomas and a Fetal Rhabdomyoma	Acta Derm Venereol. 95(2):243-4.	2015	4,24	
other	Terenziani M, D'Angelo P, Inserra A, Boldrini R, Bisogno G, Babbo GL, Conte M, Dall' Igna P, De Pasquale MD, Indolfi P, Piva L, Riccipetitoni G, Siracusa F, Spreafico F, Tamaro P, Cecchetto G	Mature and immature teratoma: A report from the second Italian pediatric study	Pediatr Blood Cancer. 62(7):1202-8.	2015	2,39	
other	Virgone C, Alaggio R, Dall'Igna P, Buffa P, Tonegatti L, Ferrari A, Bisogno G, Cecchetto G	Epithelial Tumors of the Ovary in Children and Teenagers: A Prospective Study from the Italian TREP Project	J Pediatr Adolesc Gynecol Dec;28(6):441-6	2015	1,60	

other	Chieffo DP, Tamburrini G, Caldarelli M, Di Rocco C	Preoperative neuropsychological and behavioral evaluation of children with thalamic tumors	J Neurosurg Pediatr. 13(5):507-13.	2014	1,37	
other	Stachowicz-Stencel T, Orbach D, Brecht I, Schneider D, Bien E, Synakiewicz A, Rod J, Ferrari A, Cecchetto G, Bisogno G	Thymoma and thymic carcinoma in children and adolescents: a report from the European Cooperative Study Group for Pediatric Rare Tumors (EXPeRT).	Eur J Cancer; 51(16):2444-52	2015	6,16	
other	Filippi L, Tamburini A, Berti E, Perrone A, Defilippi C, Favre C, Calvani M, Della Bona ML, la Marca G, Donzelli G	Successful Propranolol Treatment of a Kaposiform Hemangioendothelioma Apparently Resistant to Propranolol	Pediatr Blood Cancer; 63(7):1290-2	2016	2,63	
other	Cambiaso P, Amodio D, Procaccini E, Longo D, Galassi S, Camassei F, Cappa M	Pituicytoma and cushing's disease in a 7-year-old girl: A mere coincidence?	Pediatrics. 136(6):e1632-6.	2015	5,47	
Retinoblastoma	De Ioris MA, Valente P, Randisi F, Buzzonetti L, Carai A, Cozza R, Del Bufalo F, Romanzo A, Angioni A, Cacchione A, Bernardi B, Mastronuzzi A	Baseline central nervous system magnetic resonance imaging in early detection of trilateral retinoblastoma: Pitfalls in the diagnosis of pineal gland lesions	Anticancer Res. 34(12):7449-54.	2014	1,87	
Retinoblastoma	Grotta S, D'Elia G, Scavelli R, Genovese S, Surace C, Sirleto P, Cozza R, Romanzo A, De Ioris MA, Valente P, Tomaiuolo AC, Lepri FR, Franchin T, Ciocca L, Russo S, Locatelli F, Angioni A	Advantages of a next generation sequencing targeted approach for the molecular diagnosis of retinoblastoma	BMC Cancer. 15:841.	2015	3,36	
Retinoblastoma	Valente P, De Ioris MA, Romanzo A, Cozza R, Natali G, Buzzonetti L	Advanced unilateral retinoblastoma: a case of sparing enucleation treatment failure.	Can J Ophthalmol. 51(2):e40-3.	2016	1,46	
Rhabdomyosarcoma	Ciarapica R, Carcarino E, Adesso L, De Salvo M, Bracaglia G, Leoncini P, Dall'Agnese A, Verginelli F, Milano G, Boldrini R, Inserra A, Stifani S, Screpanti I, Marquez V, Valente S, Mai A, Puri P, Locatelli F, Palacios D, Rota R	Pharmacological inhibition of EZH2 as a promising differentiation therapy in embryonal RMS	BMC Cancer. 14:139.	2014	3,32	

Rhabdomyosarcoma	Conti B, Slemmons KK, Rota R, Linardic CM	Recent Insights into Notch Signaling in Embryonal Rhabdomyosarcoma	Curr Drug Targets. 17(11):1235-44.	2016	3,03	
Rhabdomyosarcoma	De Salvo M, Raimondi L, Vella S, Adesso L, Ciarapica R, Verginelli F, Pannuti A, Citti A, Boldrini R, Milano G, Cacchione A, Ferrari A, Collini P, Rosolen A, Bisogno G, Alaggio R, Inserra A, Locatelli M, Stifani S, Screpanti I, Miele L, Locatelli F, Rota R	Hyper-activation of notch3 amplifies the proliferative potential of rhabdomyosarcoma cells	PLoS One. 9(5):e96238.	2014	3,53	
Rhabdomyosarcoma	Gasparini P, Casanova M, Villa R, Collini P, Alaggio R, Zin A, Bonvini P, Antonescu CR, Boldrini R, Caserini R, Moro M, Centonze G, Meazza C, Massimino M, Bergamaschi L, Luksch R, Chiaravalli S, Bisogno G, Zaffaroni N, Daidone MG, Sozzi G, Ferrari A	Anaplastic lymphoma kinase aberrations correlate with metastatic features in pediatric rhabdomyosarcoma	Oncotarget. 7(37):58903-58914.	2016	5,01	
Rhabdomyosarcoma	Piersigilli F, Auriti C, Mondì V, Francalanci P, Salvatori G, Danhaive O	Decreased CDKN1C Expression in Congenital Alveolar Rhabdomyosarcoma Associated with Beckwith-Wiedemann Syndrome	Indian J Pediatr. 83(12-13):1476-1478.	2016	0,81	
Rhabdomyosarcoma	Vella S, Pomella S, Leoncini PP, Colletti M, Conti B, Marquez VE, Strillacci A, Roma J, Gallego S, Milano GM, Capogrossi MC, Bertaina A, Ciarapica R, Rota R	MicroRNA-101 is repressed by EZH2 and its restoration inhibits tumorigenic features in embryonal rhabdomyosarcoma	Clin Epigenetics. 7:82.	2015	4,54	
Rhabdomyosarcoma	Angelini L, Bisogno G, Alaggio R, Scarzello G, Santoro L, Zanetti I, Scagnellato A, Basso E, D'Angelo P, Ferrari A, Castagnetti M.	Prognostic factors in children undergoing salvage surgery for bladder/prostate rhabdomyosarcoma.	J Pediatr Urol. 12(4):265.e1-8	2016	1,17	

Rhabdomyosarcoma	Virgone C, Lalli E, Bisogno G, Lazzari E, Roma J, Zin A, Poli E, Cecchetto G, Dall'Igna P, Alaggio R	DAX-1 Expression in Pediatric Rhabdomyosarcomas: Another Immunohistochemical Marker Useful in the Diagnosis of Translocation Positive Alveolar Rhabdomyosarcoma	PLoS One. 10(7):e0133019	2015	3,54	
Rhabdomyosarcoma	Castagnetti M, Angelini L, Alaggio R, Scarzello G, Bisogno G, Rigamonti W	Oncologic outcome and urinary function after radical cystectomy for rhabdomyosarcoma in children: role of the orthotopic ileal neobladder based on 15-year experience at a single cente	J Urol. Jun;191(6):1850-5	2014	4,47	
Rhabdomyosarcoma	Zin A, Bertorelle R, Dall'Igna P, Manzitti C, Gambini C, Bisogno G, Rosolen A, Alaggio R	Epithelioid rhabdomyosarcoma: a clinicopathologic and molecular study	Am J Surg Pathol; 38(2):273-8	2014	5,14	
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Rhabdomyosarcoma	Tombolan L, Poli E, Martini P, Zin A, Romualdi C, Bisogno G, Lanfranchi G	NELL1, whose high expression correlates with negative outcomes, has different methylation patterns in alveolar and embryonal rhabdomyosarcoma	Oncotarget;8(20):33086-33099	2017	5,01	
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Sarcoma	Ferrari A, De Salvo GL, Brennan B, van Noesel MM, De Paoli A, Casanova M, Francotte N, Kelsey A, Alaggio R, Oberlin O, Carli M, Ben-Arush M, Bergeron C, Merks JH, Jenney M, Stevens MC, Bisogno G, Orbach D.	Synovial sarcoma in children and adolescents: the European Pediatric Soft Tissue Sarcoma Study Group prospective trial (EpSSG NRSTS 2005).	Ann Oncol;26(3):567-72.	2015	9,26	
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Pubblicazioni

Malattie ematologiche pediatriche

Unità operativa / Dipartimento /Azienda: Membri italiani pediatri dell'European Reference Network in Rare Hematological Diseases di Padova, Genova Gaslini, Firenze-Pisa, Genova Galliera, Roma Sapienza, Bari, Monza, Napoli Univ. Vanvitelli, Orbassano Torino.

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GRUPPO DI MALATTIE RARE:

Globulo Rosso (Sickle cell disease, Thalassemia);

Insufficienze midollari (Fanconi, DBA, Neutropenie);

Coagulazione (Emofilia, Malattia di Von Willebrand)

Pubblicazioni: Malattie ematologiche pediatriche

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Thalassemia	**Piga A**, Fracchia S, Lai ME, Cappelli MD, Hirschberg R, Habr D, Wegene A, Bouillaud E, **Forni GL.**	Deferasirox effect on renal haemodynamic parameters in patients with transfusion-dependent β thalassaemia.	Br J Haematol. 2015 Mar;168(6):882-90. doi: 10.1111/bjh.13217.	2014	5.401	
Thalassemia	**Piga A,** Longo F, Origa R, Roggero S Pinna F, Zappu A, Castiglioni C, Cappellini MD.	Deferasirox for cardiac siderosis β-thalassaemia major: a multicentre, open label, prospective study.	Br J Haematol. 2014 Nov;167(3):423-6. doi: 10.1111/bjh.12987	2014	5.401	
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Thalassemia	**Pepe A**, Rossi G, Bentley A, Putti MC, Frizziero L, D'Ascola DG, Cuccia L, Spasiano A, Filosa A, Caruso V, Ha A, Meloni A.	Cost-Utility Analysis of Three Iron Chelators Used in Monotherapy for the Treatment of Chronic Iron Overload in β-Thalassaemia Major Patients: An Italian Perspective.	Clin Drug Investig. 7(5):453-464	2017	1.806	
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Thalassemia	Meloni A, Restaino G, Missere M, De Marchi D, Positano V, Valeri G, Giuseppe D'Ascola D, Peluso A, Caterina Putti M, Lendini M, Giovan Neri M, Midiri M, Sallustio G, **Pepe A**	Pancreatic iron overload by T2* M in a large cohort of well treated thalassemia major patients: can it tell us heart iron distribution and function?	Am J Hematol. 90(9):E189-90.	2015	5.000	
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Talassemia	Zebisch A, Schulz E, Grosso M, Lombardo B, Acierno G, Sill H, **Iolascon A.**	Identification of a novel variant of epsilon-gamma-delta-beta thalassemia highlights limitations of next generation sequencing	Am J Hematol. 2015 Mar;90(3):E52-4. doi: 10.1002/ajh.23913	2015	5.000	
Sferocitosi Ereditaria	Farruggia P, Puccio G, Ramenghi U, Colombatti R, Corti P, Trizzino A, Barone A, Boscarol G, Ferraro F, Gro P, Lo Valvo L, Luti L, Matarese SMR, Mosa C, Putti MC, Rubert L, Ruffo G Sainati L, Tartaglione I, Russo G, **Perrotta S.**	Recombinant erythropoietin vs. blood transfusion care in infants with hereditary spherocytosis: a retrospective cohort study of A.I.E.O.P. patients (Associazione Italiana Emato-Oncologia Pediatrica).	Am J Hematol. 2017 Jun;92(6):E103-E105.	2017	5.000	

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Pseudoiperkaliemia familiare	Andolfo I, Russo R, Manna F, De Rosa G, Gambale A, Zouwail S, Detta N, Pardo CL, Alper SL, Brugnara C, Sharma AK, De Franceschi L, **Iolascon A.**	Functional characterization of novel ABCB6 mutations and their clinical implications in familial pseudohyperkalemia.	Haematologica. 2016 Aug;101(8):909-17. doi: 10.3324/haematol.2016.142372.	2016	6.671	
Globulo rosso (NGS)	Russo R, Andolfo I, **Iolascon A**.	Next generation research and therapy in red blood cell diseases	Haematologica. 2016 May;101(5):515-7. doi: 10.3324/haematol.2015.139238	2016	6.671	
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Anemia Aplastica	**Dufour C**, Pillon M, Sociè G, Rovò A, Carraro E, Bacigalupo A, Oneto R, Passweg J, Risitano A, Tichelli A, Peffault de Latour R, Schrezenmeier Hocshmann B, Peters C, Kulasekarar A, Van Biezen A, Samarasinghe S, Hussein AA, Ayas M, Aljurf M, Marsh	Outcome of aplastic anaemia in children. A study by the severe aplastic anaemia and paediatric disease working parties of the European group blood and bone marrow transplant.	Br J Haematol. 2015 May;169(4):565-73.	2014	5.401	
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Anemia di Fanconi	**Dufour Carlo**	How I manage patients with Fanc anaemia	BJ Haematology 10.1111/bjh.14615	2017	5.401	

Anemia Aplastica	**Miano M**, Lanciotti M, Giardino S, Dufour C.	Ser245Tyr TINF2 mutation in a lo term survivor after a second myeloablative SCT following late graft failure for Aplastic Anaemia	Blood Cells Mol Dis. 2015 Aug;55(2):187-8. d 10.1016/j.bcmd.2015.01.002	2015	2.731	
Anemia Aplastica	**Dufour C**, Pillon M, Passweg J, Socié Bacigalupo A, Franceschetto G, Carr E, Oneto R, Risitano AM, Peffault de Latour R, Tichelli A, Rovo A, Peters C Hoechsmann B, Samarasinghe S, Kulasekararaj AG, Schrezenmeier H, Aljurf M, Marsh J.	Outcome of aplastic anemia in adolescence: a survey of the Seve Aplastic Anemia Working Party of the European Group for Blood an Marrow Transplantation.	Haematologica. 2014 Oct;99(10):1574-81. do 10.3324/haematol.2014.106096. Epub 2014 Aug 1	2014	6.671	
Anemia di Fanconi	Svahn J, Bagnasco F, Cappelli E, Onofrillo D, Caruso S, Corsolini F, De Rocco D, Savoia A, Longoni D, Pillon Marra N, Ramenghi U, Farruggia P, Locasciulli A, Addari C, Cerri C, Mastrodicasa E, Casazza G, Verzegn F, Riccardi F, Haupt R, Barone A, Ces S, Cugno C **, Dufour C**	Somatic, hematologic phenotype, long-term outcome, and effect of hematopoietic stem cell transplantation. An analysis of 97 Fanconi anemia patients from the Italian national database on beha of the Marrow Failure Study Grou of the AIEOP (Italian Association Pediatric Hematology-Oncology).	Am J Hematology, 91(7):666-71. doi: 10.1002/ajh.24373	2016	5.000	
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Anemia di Fanconi	Svahn J, Lanza T, Rathbun K, Bagby Ravera S, Corsolini F, Pistorio A, Longoni D, Farruggia P, Dufour C, **Cappelli E**	p38 Mitogen-activated protein kinase inhibition enhances in vitr erythropoiesis of Fanconi anemia complementation group A-deficie bone marrow cells.	Exp Hematol. 2015 Apr;43(4):295-9. doi: 10.1016/j.exphem.2014.11.010.	2015	2.820	
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Coagulazione (Emofilia	Castaman G, Rocino A, Mazzucconi MG, Zanon E, Gagliano F, **Molinari A**	Prophylaxis therapy in paediatric patients with haemophilia: a surv of clinical management trends in Italy.	Blood Transfus. 2015 Oct;13(4):631-8	2015	1.514	
Coagulazione	**Giordano P**, Francavilla M, Buonamico P, Suppressa P, Lastella P, Sangerardi M, Miniello VL, Scardapane A, Lenato GM, Sabbà C.	Hepatic angiodynamic profile in paediatric patients with hereditar haemorrhagic telangiectasia type and type 2.	Vasa. May;46(3):195-202.	2017	1.122	
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Coagulazione (Emofilia	**Giordano P**, Lassandro G, Valente M, Molinari AC, Ieranò P, Coppola A.	Current management of the hemophilic child: a demanding interlocutor. Quality of life and adequate cost-efficacy analysis.	Pediatr Hematol Oncol. Nov;31(8):687-702.	2014	1.090	

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Trial clinici farmacologici

Lo stato dell'arte sullo sviluppo dei farmaci per malattie rare, frutto sia della ricerca industriale che indipendente, con la partecipazione alle sperimentazioni di gruppi italiani: i dati dell'Associazione Italiana di Ematologia ed Oncologia Pediatrica (AIEOP)

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Ogni anno in Europa a 15.000 bambini di età compresa tra 0-14 anni e a 20.000 adolescenti e giovani adulti di età compresa tra i 15 ei 24 anni viene diagnosticato un tumore [1]. La sopravvivenza complessiva a 5 anni è continuamente migliorata e si è giunti a quasi l'80% subito dopo gli anni 2000 [2]. Ciò vuol dire che comunque 6.000 giovani in Europa muoiono ancora di tumore ogni anno, essendo i tumori la prima causa di morte in questa categoria [3].

In Italia vengono effettuate ogni anno circa 1.200 nuove diagnosi di tumore nell'età pediatrica (0-18 anni), la quasi totalità presso una delle istituzioni dell'AIEOP. Fin dalla sua fondazione l'AIEOP ha costituito il punto di riferimento nazionale per la cura delle malattie oncologiche, emato-oncologiche ed ematologiche dell'infanzia. Nel tempo l'AIEOP ha progressivamente intrapreso la strada delle sperimentazioni cliniche controllate su scala nazionale, favorendo così la crescita di una generazione di ricercatori di alto profilo scientifico nelle diverse istituzioni che la compongono. L'AIEOP si è anche fatta carico negli ultimi due-tre decenni di fare da sponsor di molti trials clinici con una progressiva assunzione di responsabilità scientifiche, organizzative e normative negli ambiti di propria pertinenza, contribuendo altresì alla sistematizzazione degli assetti relativi alla centralizzazione delle diagnosi, alla gestione dei dati clinici e del materiale biologico centralizzato.

In relazione agli Early Clinical Trials (ECT, studi di fase I-II con farmaci non ancora registrati per l'età pediatrica) alcuni dei più importanti centri della rete AIEOP partecipano attivamente agli studi promossi dal network europeo ITCC (Innovative Therapies for Children with Cancer), che riunisce oltre 50 istituzioni europee in un consorzio di eccellenza di vocazione prettamente accademica (orientato quindi a studi no-profit) ma che contribuisce anche alla implementazione e gestione di ECTs parzialmente sponsorizzati dall'industria farmaceutica. Gli obiettivi dell'ITCC sono quelli di identificare nuove molecole potenzialmente utilizzabili nell'impiego clinico, di accreditare centri europei di eccellenza che possano arruolare pazienti in studi clinici di fase I-II e di garantire che la conduzione di tali studi avvenga in accordo con principi di Good Clinical Practice (GCP).

In base al rapporto nazionale 2016 (sperimentazioni relative all'anno 2015) [4] in Italia sono state condotte 98 sperimentazioni cliniche sui minori di 18 anni, di cui 54 esclusivamente su pazienti pediatrici e 44 su minori e adulti. Le aree terapeutiche più rappresentate erano la neurologia/psichiatria, la infettivologia, l'area cardio-vascolare e l'oncologia/oncoematologia. Vale la pena sottolineare che queste ultime sperimentazioni vengono esclusivamente condotte presso centri dell'AIEOP.

Allo scopo di favorire la collaborazione tra i vari centri della rete AIEOP e soprattutto per garantire ad ogni paziente la possibilità di accedere alle terapie più innovative per le varie fasi della sua malattia, soprattutto quelle relative alle malattie resistente o in recidiva, il Gruppo di Lavoro di Farmacologia Clinica e Farmacosorveglianza dell'AIEOP ha creato un registro degli studi clinici con nuovi farmaci attivi nei centri AIEOP. Il

registro degli studi è stato collocato all'interno del sito web dell'AIEOP e reso interamente fruibile da parte di tutti i soci, così che essi possano essere messi facilmente a conoscenza di eventuali studi aperti in altri centri e di poter quindi avere a portata di "click" i contatti per l'acquisizione delle informazioni principali. Si tratta di 26 studi dei quali 10 in oncologia, 11 in oncoematologia e 5 in altre aree di pertinenza alle patologie gestite nei entri AIEOP. In questi studi sono sperimentati 24 farmaci innovativi e sono coinvolti 17 centri AIEOP. Uno studio è nazionale e 25 sono internazionali. Sono stati inoltre individuate 3 studi internazionali con altrettanti nuovi farmaci, la cui approvazione in Italia è prevista nei prossimi mesi.

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Trial clinici farmacologici: Malattie oncologiche

1 Nome MR	2 *Sponsor*	3 *Partners*		4 Ruolo		5 Denominazione del *trial*	6 Farmaco	7 Durata		8 Note
		I	N	C	P			C	T (anno)	
Leucemia Linfoblastica Acuta	AMGEN	X				An Open-Label, Multi-center, Expanded Access Protocol of Blinatumomab for the Treatment of Pediatric and Adolescent Subjects with Relapsed and/or Refractory B-precursor Acute Lymphoblastic Leukemia (ALL) (Rialto Study)	Blinatumomab	X		Centro coordinatore: Ospedale Pediatrico Bambino Gesù, Roma Centri partecipanti: Ospedale San Gerardo, Monza
Leucemia Linfoblastica Acuta	AMGEN	X				A Randomized, Open-label, Controlled Phase 3 Adaptive Trial to Investigate the Efficacy, Safety, and Tolerability of the BiTE Antibody Blinatumomab as Consolidation Therapy Versus Conventional Consolidation Chemotherapy in Pediatric Subjects With High-risk First Relapse B-precursor Acute Lymphoblastic Leukemia (ALL)	Blinatumomab	X		Centro coordinatore: Ospedale Pediatrico Bambino Gesù, Roma Centri partecipanti: -Ospedale San Gerardo, Monza - Policlinico S. Orsola-Malpighi , Bologna - IRCCS Istituto Giannina Gaslini , Genova - Ospedale Santobono Pausilipon, Napoli - Azienda Ospedaliero-

										Universitaria di Padova - Policlinico San Matteo, Pavia - Policlinico Umberto I - Roma
Leucemia Linfoblastica Acuta B	NOVARTIS	X				A Phase II, single arm, multicenter trial to determine the efficacy and safety of CTL019 in pediatric patients with relapsed and refractory B-cell acute lymphoblastic leukemia	CTL019 chimeric antigen receptor T cell (CART)	X		Centro coordinatore: Ospedale San Gerardo (Monza)
Leucemia Linfoblastica Acuta	AMGEN	X				Phase 1b/2 Study of Carfilzomib in Combination with Dexamethasone, Mitoxantrone, PEG-asparaginase, and Vincristine (UK R3 Induction Backbone) in Children with Relapsed or Refractory Acute Lymphoblastic Leukemia-CFZ008	Carfilzomib	X		Centro coordinatore: Ospedale San Gerardo (Monza) Centri partecipanti: - Ospedale Pediatrico Bambino Gesù, Roma
Leucemia Acuta Mieloide	Boehringer Ingelheim	X				Open-label, dose-escalating trial to evaluate the tolerability, toxicity, safety, pharmacokinetics, pharmacodynamics and activity of volasertib added to the standard intensive salvage chemotherapy regimen with liposomal daunorubicine,	Volasertib	X		Centro coordinatore: Ospedale Pediatrico Bambino Gesù, Roma Centri partecipanti: Ospedale San Gerardo, Monza

						fludarabine and cytarabine (DNX-FLA) followed by fludarabine and cytarabine (FLA) in children from 3 months to less than18 years of age with acute myeloid leukaemia after failure of the front-line therapy Eudract: 2015-004625-14				
Sindrome mielodisplastica e Leucemia mielomonocitica giovanile	CELGENE	X				A Phase 2, Multicenter, Open-Label Study To Evaluate The Pharmacokinetics, Pharmacodynamics, Safety And Activity Of Azacitidine And To Compare Azacitidine To Historical Controls In Pediatric Subjects With Newly Diagnosed Advanced Myelodysplastic Syndrome Or Juvenile Myelomonocytic Leukemia Before Hematopoietic Stem Cell Transplantation Numero Eudract: 2014-002388-13	Azacitidina	X		Centro coordinatore: Ospedale Pediatrico Bambino Gesù, Roma Centri partecipanti: -Ospedale San Gerardo, Monza - Azienda Ospedaliero-Universitaria di Padova - Policlinico San Matteo (Pavia) - Policlinico S. Orsola-Malpighi (Bologna) - IRCCS Istituto Giannina Gaslini (Genova) - Ospedale Regina Margherita - Sant'Anna di Torino

Sindrome mielodisplastica e Leucemia mielomonocitica giovanile	CELGENE	X				A phase I/II study of Azacitidine (Vidaza) in pediatric patients with newly diagnosed or relpased high-grade pediatric MDS or JMML Eudract: 2010-022235-10	Azacitidina	X		Centro coordinatore: Osp. San Gerardo (Monza) Centri partecipanti: - Osp. Pediatrico Bambino Gesù - Policlinico San Matteo (Pavia)
Linfoma a larghe cellule anaplastiche + Linfoma di Hodgkin	Millennium Pharmaceuticals	X				A phase I/II study of brentuximab vedotin in pediatric patients with relapsed or refractory sistemic anaplastic large-cell lymphoma or hodgkin lymphoma Eudract: 2011-001240-29	Brentuximab	X		Centro coordinatore: Azienda Ospedaliero-Universitaria di Padova Centri partecipanti: Ospedale Pediatrico Bambino Gesù, Roma
Linfoma non-Hodgkin	Janssen	X				A Randomized, Open-label, Safety and Efficacy Study of Ibrutinib in Pediatric and Young Adult Patients With Relapsed or Refractory Mature B-cell non-Hodgkin Lymphoma. EudraCT: 2016-000259-28	Ibrutinib	X		Centro coordinatore: Ospedale Pediatrico Bambino Gesù, Roma Centri partecipanti: - Ospedale Regina Margherita - Sant'Anna di Torino -Ospedale San Gerardo, Monza - Azienda Ospedaliero-Universitaria di Padova

Linfoistiocitosi emofagocitica primitiva	Novimmune SA	X				A Pilot, Open-label, Single Arm, Multicentre Study to Explore Safety, Tolerability, Pharmacokinetics and Efficacy of Intravenous Multiple Administrations of NI-0501, an Anti-interferon Gamma (Anti-IFNγ) Monoclonal Antibody, in Paediatric Patients with Primary Haemophagocytic Lymphohistiocytosis in whom the disease has reactivated or an unsatisfactory response has been achieved	Anti-IFNγ	X		Centro coordinatore: Ospedale Pediatrico Bambino Gesù, Roma Centri partecipanti: - Ospedale San Gerardo (Monza) - Ospedale Meyer (Firenze); - Ospedale Gaslini, (Genova); - Ospedale Pausilipon (Napoli); - Azienda Ospedaliero-Universitaria di Padova; - Ospedale Regina Margherita - Sant'Anna di Torino
Neuroblastoma	University of Birmingham	X				Studio randomizzato di fase IIb su Bevacizumab aggiunto a temozolomide +/- Irinotecan in bambini con neuroblastoma refrattario/recidivato	Bevacizumab	X		Centro coordinatore: Ospedale Pediatrico Bambino Gesù, Roma Centri partecipanti: - Istituto Nazionale dei Tumori di Milano - Istituto Gianna Gaslini, Genova

Neuroblastoma	St. Anna Kinderkrebsforschung	X				A phase I/II dose schedule finding study of CH14.18/CHO continuous infusion combined with subcutaneous aldesleukin (IL-2) in patients with primary refractory or relapsed neuroblastoma.	CH14.18/CHO	X		Centro coordinatore: Istituto Gianna Gaslini, Genova Centri partecipanti: - Istituto Nazionale dei Tumori di Milano - Ospedale Pediatrico Bambino Gesù, Roma
Tumori neuroectodermici, rabdomiosarcoma, tumori solidi con deregolazione via ErbB	Boehringer Ingelheim	X				Studio di fase I in aperto, a dosaggio scalare, per determinare la MTD, la sicurezza, la PK e l'efficacia di afatinib in monoterapia nei bambini di età compresa tra 2 anni e <18 anni con tumori recidivanti/refrattari neuroectodermici, rabdomiosarcoma e/o altri tumori solidi con nota deregolazione della via ErbB indipendentemente dalla istologia tumorale Eudract: 2014-002123-10	Afatinib	X		Centro coordinatore: Ospedale Pediatrico Bambino Gesù, Roma Centri partecipanti: - Istituto Nazionale dei Tumori di Milano - Istituto Gianna Gaslini, Genova - Azienda Ospedaliero-Universitaria di Padova
Melanoma + tumori solidi e linfomi PD-L1+	Merck sharp & dohme corp	X				A phase 1/2 study of Pembrolizumab (MK-3475) in children with advanced melanoma or a PD-L1 positive advanced, relapsed or refractory solid tumor or lymphoma (KEYNOTE-051) Eudract: 2014-002950-38	Pembrolizumab	X		Centro coordinatore: Ospedale Pediatrico Bambino Gesù, Roma

Tumori solidi positivi per mut BRAF V600	GlaxoSmithKline	X				Studio multicentrico di fase 1/2a, a braccio singolo, in aperto, in 2 parti per determinare la sicurezza, la tollerabilità e la farmacocinetica di dabrafenib orale in bambini e adolescenti con tumori solidi in stadio avanzato positivi alla mutazione del gene BRAF V600. Eudract: 2012-001499-12	Dabrafenib	X		Centro coordinatore: Istituto Nazionale dei Tumori di Milano
Tumori con alterazioni di ALK	Novartis	X				Studio di Fase I, in aperto, di incremento della dose di LDK378, in pazienti pediatrici con tumori caratterizzati da alterazioni genetiche della chinasi del linfoma anaplastico (ALK) Numero EUDRACT: 2012-002074-31	LDK378 (Ceritinib)	X		Centro coordinatore: Istituto Nazionale dei Tumori di Milano

Tumori solidi	Roche	X				Studio Di Fase Iniziale, Multicentrico, In Aperto, Sulla Sicurezza E La Farmacocinetica Dell'anticorpo Anti-Pd-L1 (Mpdl3280a) In Pazienti Pediatrici E Giovani Adulti Con Tumori Solidi Precedentemente Trattati" Numero Eudract : 2014-004697-41	Atezolizumab	X		Centro coordinatore: Istituto Nazionale dei Tumori di Milano Centri partecipanti: - Azienda Ospedaliero Universitaria di Padova - Azienda Ospedaliera Universitaria di Torino - IRCCS Istituto Giannina Gaslini (Genova) - Ospedale Pediatrico Bambino Gesù (Roma) - Policlinico Gemelli - Roma - Azienda Ospedaliera San Gerardo (Monza)
Tumori solidi	Celgene	X				"Studio di fase 1/2, multicentrico, in aperto, di definizione della dose, per una valutazione della sicurezza e della tollerabilità e una valutazione preliminare dell'efficacia di nab®-paclitaxel somministrato settimanalmente in soggetti in età pediatrica con tumori solidi recidivati o refrattari" EUDRACT: 2013-000144-26	nab-paclitaxel	X		Centro coordinatore: Istituto Nazionale dei Tumori di Milano Centri partecipanti: -Ospedale Meyer (Firenze) -IRCCS Istituto Giannina Gaslini (Genova) -Azienda Ospedaliero Universitaria di Padova

										-Ospedale Pediatrico Bambino Gesù Roma - Azienda Ospedaliera Universitaria di Torino
Tumori solidi	Roche	X				Studio di FASE I/II, multicentrico, in aperto, a incremento progressivo della dose, sulla sicurezza e la farmacocinetica di Cobimetinib in pazienti pediatrici e giovani adulti con tumori solidi precedentemente trattati EUDRACT : 2014-004685-25	Cobimetinib	X		Centro coordinatore: Istituto Nazionale dei Tumori di Milano Centri partecipanti: - Azienda Ospedaliero Universitaria di Padova - Azienda Ospedaliera Universitaria di Torino - IRCCS Istituto Giannina Gaslini (Genova) - Ospedale Pediatrico Bambino Gesù (Roma) - Policlinico Gemelli – Roma
Tumori solidi	Eisai Co., Ltd.	X				Phase 1/2 study of Lenvatinib in children an adolescent with relapsed or refractory solid malignancies Eudract: 2013-005534-38	Lenvatinib	X		Centro coordinatore: Ospedale Pediatrico Bambino Gesù, Roma Centri partecipanti: - Istituto Nazionale dei Tumori di Milano

Anemia falciforme	AstraZeneca	X				Multicenter, open-label, randomised, pharmacokinetic (PK) and pharmacodynamic (PD) doseranging Phase II study of ticagrelor followed by a double-blind, randomized, parallel-group, placebo-controlled 4 weeks extension phase in pediatric patients with sickle cell disease AstraZeneca Protocol D5136C00007 Eudract 2014-001006-18	Tigacleror	X		Centro coordinatore: Ospedale Pediatrico Bambino Gesù, Roma
Graft-Versus-Host Disease	Ospedale Pediatrico Bambino Gesù, Roma		X			Studio di fase II, multicentrico, randomizzato, controllato in aperto sull'impiego di siero antilinfocitario di coniglio e rituximab per la modulazione della malattia del trapianto contro l'ospite nel contesto del trapianto allogenico di cellule staminali emopoietiche da donatore HLA compatibile in pazienti affetti da patologie non maligne EudraCT: 2011-004730-34	ATG-Fresenius Rituximab	X		Centro coordinatore: Ospedale Pediatrico Bambino Gesù, Roma Centri partecipanti: - Ospedale San Gerardo (Monza) - Istituto San Raffaele di Milano - Ospedale Binaghi di Cagliari

Trombosi venose	Pfizer	X				A randomized, open-label, active controlled, safety and extrapolated efficacy study in pediatric patients requiring anticoagulation for the treatment of a venous thromboembolic event B0661037/CV185-325 Eudract: 2014-002606-20	Apixaban	X		Centro coordinatore: Ospedale Pediatrico Bambino Gesù, Roma
Tromboembolia venosa	Boehringer Ingelheim	X				Studio in aperto, multicentrico, randomizzato, con controllo attivo, per gruppi paralleli, di non inferiorità, con dabigatran etexilato verso una terapia standard per il trattamento della tromboembolia venosa in bambini dalla nascita fino ai 18 anni: studio DIVERSITY. EudraCT: 2013-002114-12	Dabigatran	X		Centro coordinatore: Ospedale Pediatrico Bambino Gesù, Roma Centri partecipanti: -IRCCS Fondazione Ospedale Maggiore Policlinico di Milano - Università "La Sapienza" di Roma - Ospedale Regina Margherita - Sant'Anna di Torino - Policlinico P. Giaccone di Palermo
Tromboembolia venosa	Boehriger Ingelheim	X				Studio prospettico, in aperto, con dabigatran etexilato, per la prevenzione secondaria della tromboembolia venosa in bambini dalla nascita fino ai 18 anni.	Dabigatran	X		Centro coordinatore: Ospedale Pediatrico Bambino Gesù, Roma Centri partecipanti: - Università "La Sapienza" di Roma - Ospedale Regina Margherita - Sant'Anna di Torino - Policlinico P. Giaccone di Palermo

Emorragia da Dabigatran	AstraZeneca AB	X				Studio clinico in aperto, non controllato, per valutare la sicurezza di una singola dose di idarucizumab, somminstrato per via endovenosa in pazienti pediatrici arruolati negli studi in corso di fase IIb/III con dabigatran etexilato per il trattamento e la prevenzione secondaria del tromboembolismo venoso.	Idarucizumab	X		Centro coordinatore: Ospedale Pediatrico Bambino Gesù, Roma Centri partecipanti: -IRCCS Fondazione Ospedale Maggiore Policlinico di Milano - Università "La Sapienza" di Roma - Ospedale Regina Margherita - Sant'Anna di Torino - Policlinico P. Giaccone di Palermo
Leucemia Mieloide cronica	Erasmus MC	X				A phase I/II study of Bosutinib in pediatric patients with Chronic Myeloid Leukemia who are resistant or intolerant to at least one prior Tyrosine Kinase Inhibitor therapy, ITCC-054/AAML1621 EudraCT: 2015-002916-34	Bosutinib	X		In sottomissione in Italia
Tumori pediatrici con alterazione di ALK, ROS1, MET	Erasmus MC	X				A phase 1B of crizotinib either in combination or as single agent in pediatric patients with ALK, ROS1 or MET positive malignancies Study ITCC 053 EudraCT: 2015-005437-53	Crizotinib	X		In sottomissione in Italia

Leucemia Linfoblastica Acuta CD22+	Erasmus MC	X				A phase I/II study of Inotuzumab Ozogamicin as a single agent and in combination with chemotherapy for pediatric CD22-positive relapsed/refractory Acute Lymphoblastic Leukemia EudraCT: 2016-000227-71	Inotuzumab Ozogamicin	X		In sottomissione in Italia

Trial clinici farmacologici

Malattie ematologiche

Unità operativa / Dipartimento /Azienda: Membri italiani pediatri dell' European Reference Network in Rare Hematological Diseases di Padova, Genova Gaslini, Firenze-Pisa, Genova Galliera, Roma Sapienza, Bari, Monza, Napoli Univ. Vanvitelli, Orbassano Torino.

Scheda compilata da: Raffaella Colombatti (Padova), Carlo Dufour (Genova Gaslini), Claudio Favre (Firenze), Gianluca Forni (Genova Galliera), Fiorina Giona (Roma Sapienza), Paola Giordano (Bari), Nicoletta Masera (Monza), Silverio Perrotta (Napoli Univ. Vanvitelli), Antonio Piga (Torino).

GRUPPO DI MALATTIE RARE:

Globulo Rosso (Sickle cell disease, Thalassemia);

Insufficienze midollari (Fanconi, DBA, Neutropenie);

Coagulazione (Emofilia, Malattia di Von Willebrand)

Trial clinici farmacologici: Malattie ematologiche

1 **Nome MR**	2 ***Sponsor***	3 ***Partners***		4 **Ruolo**		5 **Denominazione del *trial***	6 **Farmaco**	7 **Durata**		8 **Note**
		I	**N**	**C**	**P**			**C**	**T (anno)**	
Sickle cell disease	Eli Lilly	**X**			**X**	H7T-MC-TADO, DOVE Trial: A Phase 3, Double-Blind, Randomized, Efficacy and Safety Comparison of Prasugrel and Placebo in Pediatric Patients With Sickle Cell Disease. NCT01794000	**Prasugrel**		**2015**	**Centri partecipanti: Padova**, Genova Galliera, Monza, Modena, Verona
Sickle cell disease	AddMedica	**X**			**X**	ESCORT_HU Trial: European Sickle Cell Disease Cohort – Hydroxyurea. NCT02516579	**Idrossiurea**	**X**		**Centro coordinatore:** Padova **Centri partecipanti:** Napoli Univ Vanvitelli, Modena, Verona

Sickle cell disease	Global Blood Therapeutics	**X**			**X**	HOPE: A Phase 3, Double-blind, Randomized, Placebo-controlled, Multicenter Study of GBT440 Administered Orally to Patients with Sickle Cell Disease	**GBT440-131**	**X**		**Centro partecipante:** Padova, Genova Galliera
Sickle cell disease	Pfizer		**x**		**x**	PCV13 vaccine in children 6-17 years with SCD	**PCV13**		**2014**	**Phase 3** **Centro partecipante:** Torino
Thalassemia major	Celgene	**x**		**x**		**Ace 536-BTHAL001** A Phase 3, double-blind, randomized, placebo-controlled, multicenter study to determine the efficacy and safety of Lustatercept (ACE – 536) versus placebo in adults who require regular red blood cell transfusion due to Beta – Thalassemia	**Luspatercept**	**x**		**Phase 3** **Centro coordinatore:** Torino **Centri partecipanti:** Napoli Univ Vanvitelli, Genova Galliera

Thalassemia	Acceleron	**x**		**x**		**Ace 536-06** ACE-536 Extension Study - Beta Thalassemia	**Luspatercept**	**X**		**Phase 2** **Centro coordinatore:** Torino **Centri partecipanti:** Napoli Univ Vanvitelli
Thalassemia	Acceleron	**x**		**x**		**Ace 536-04** Study to Evaluate the Effects of ACE-536 in Patients With Beta-thalassemia.	**Luspatercept**	**X**		**Phase 2** **Centro coordinatore:** Torino **Centri partecipanti:** Napoli Univ Vanvitelli, Genova Galliera
Thalassemia	Celgene	**x**		**x**		**Ace 11-BTHAL001** A *P*hase 2A, Open-Label, dose finding study to determine the safety and tolerability of sotatercept (ACE-011) in adults with beta (ß)-thalassemia".	**Sotatercept**	**x**		**Phase 2** **Centri partecipanti:** Genova Galliera

Transfusion dependent anemia	Novartis	**x**			**x**	**CICL670F2202** A randomized, open label, multicenter, two arm, phase II study to evaluate treatment compliance, efficacy and safety, of an improved, deferasirox formulation (granules) in pediatric patients with iron overload	**DFX formulazione granulare**	**X**		**Phase 2** **Centro coordinatore:** Genova Galliera **Centri partecipanti:** Torino
Thalassemia major	Cerus	**x**		**x**		**CL00076**	**S-303**		**X (14.09.2015)**	**Phase 2** **Centri partecipanti:** Torino
Thalassemia major	Shire_Ferrokin	**x**			**x**	**SPD602-301 (FBS0701-CTP-15)** Studio multicentrico per monitorare il potenziale sviluppo di tumori renali in soggetti con sovraccarico di ferro trasfusionale precedentemente esposti a Deferitazole	**FB50701**		**X (2015)**	**Phase 3** **Centri partecipanti:** Torino, Genova Galliera

Thalassemia major	Novartis	**x**		**x**	**x**	**CICL670F2201** A randomized, open-label, multicenter, two arm, phase II study to investigate the benefits of an improved deferasirox formulation (film-coated tablet) –	**DFX formulazione FCT**		**X (2016)**	**Phase 2** **Centro coordinatore:** Napoli Univ Vanv
Thalassemia major	Novartis	**x**		**x**	**x**	**CICL670AIC04** Extended Evaluation of Deferasirox Film-Coated Tablet (FCT) Formulation	**DFX formulazione FCT**	**X**		**Phase 3** **Centro coordinatore:** Napoli Univ Vanv
Thalassemia major	Novartis		**x**	**x**		**DFX on renal haemodynamic parameters**	**DFX**		**X (2014)**	**Centro coordinatore:** Torino **Centri partecipanti:** Genova Galliera
Thalassemia major			**x**		**x**	**Use of amlodipine in addition to standard chelation**	**Amlodipine**		**X (2016)**	**Phase 2** **Centro coordinatore:** Torino

Thalassemia major	Novartis		**X**		**X**	**CICL670AIT14** A retrospective data collection study to assess the long term renal safety of deferasirox in patients with transfusional hemosiderosis who were enrolled into the registration studies"	**DFX**		**X (2016)**	**Centri partecipanti:** Torino, Napoli Univ, Genova Galliera
Thalassemia major	F.Hoffmann-La Roche Ltd	**X**		**X**	**X**	BP39642 " A phase II, single arm, multicenter, proof-of-mechanism study to investigate the safety, tolerability, efficacy, pharacokinetics, and pharmacodynamics of Bitopertin (RO4917838) in adults with non-transfusion-dependent B-Thalassemia	**Bitopertin**	**X**		**Phase 2 Centri partecipanti:** Genova Galliera, Napoli Univ Vanv
Talassemia Major/Emosiderosi trasfusionale	Fondazione F. e P. Cutino Onlus		**x**		**x**	**Trial sequenziale**	**Deferasirox/Deferiprone**	**C**		**Centrio partecipante:** Monza
Atansferrinemia	Sanquin	**x**		**x**		**MD2009.04**	**Apotransferrina**	**C**		**Centrio partecipante:** Monza

Thalassemia major	CVBF- CONSORZIO PER VALUTAZIONI BIOLOGICHE E FARMACOLOGICHE	**x**			**x**	**DEEP-2:** Multicentre, randomised, open label, non-inferiority active-controlled trial to evaluate the efficacy and safety of deferiprone compared to deferasirox in paediatric patients aged from 1 month to less than 18 years of age affected by transfusion-dependent haemoglobinopathies	deferiprone	x		**Centrio partecipante:** Firenze, padova
Thalassemia major	UKER-Universitätsklinikum Erlangen	**x**			**x**	**DEEP 3:** Long-term observational safety study to evaluate the nature and incidence of adverse effects of deferiprone treatment in patients with beta-thalassaemia major aged from 1 month to less than 18 years	Deferiprone	x		**Centro partecipante:** Firenze, Padova

ANEMIA DI FANCONI	Istituto G. Gaslini	**X**			**X**	Database Nazionale Osservazionale Anemia di Fanconi		**C**		**Centri partecipanti:** Genova Gaslini , Napoli Univ Vanv, Padova, Monza,
APLASIA MIDOLLARE ACQUISITA	EBMT	**X**			**X**	RACE. Studio prospettico randomizzato fra IS clasica (ATG+CsA) e IS + Eltrombopag	Eltrombopag			**Centro partecipanti:** Genova Gaslini
Coagulopatia	Boehringer Ingelheim	**X**			**X**	**1160.106**	**Dabigatran exilate**	**X**		**Centri partecipanti:** Genova gaslini, Roma Sapienza
Malattia di Gaucher	AOU Policlinico S.Orsola-Malpighi Bologna		**X**		**X**	**GAU-PED**	**-**	**X**		**Centri partecipanti:** Genova, Napoli Univ Vanv, Roma sapienza

Coagulazione-Emofilia	Baxalta US	**X**			**X**	BAX 326. A Phase 2/3 Prospective, Uncontrolled, Multicenter Study Evaluating Pharmacokinetics, Efficacy, Safety, and Immunogenicity in Previously Treated Pediatric Patients With Severe (FIX Level < 1%) or Moderately Severe (FIX Level 1-2%) Hemophilia B	**Recombinant factor IX**		**X (2016)**	**Centro partecipante:** Padova
Coagulazione-Emofilia	Bayer	**X**			**X**	BAY81-8973/13400 A multi center phase III uncontrolled open label trial to evaluate safety and efficacy of BAY81-8973 in children with severe hemophilia A under prophylaxis therapy	**Recombinant Factor VIII (BAY81-8973)**	**X**		**Centro partecipante:** Padova
Coagulazione-Von Willebrand		**X**			**X**	PROWILL: Efficacy, safety and pharmaeconomic assessment of secondary long-term prophylaxis with higly purified standardized doubly virus inactivate FVIII/FvW concentrates in patients with severe inherited VWD and frequent bleeding		**X**		**Centro partecipante:** Padova

Coagulazione (Emofilia)	Baxalta US	**X**			**X**	BAX 326 (rFIX) Continuation Study. BAX 326 (Recombinant Factor IX): Evaluation of Safety, Immunogenicity, and Hemostatic Efficacy in Previously Treated Patients With Severe (FIX Level < 1%) or Moderately Severe (FIX Level <= 2%) Hemophilia B - A Continuation Study	**Recombinant Factr IX**	**X**		**Centro partecipante:** Padova

Partecipazione Nazionale a ERN

1. La ricognizione dei centri pediatrici che sono stati accettati nella rete europea di riferimento (ERN) per le malattie emato-oncologiche pediatriche

Franca Fagioli
Direttore SC Oncoematologia Pediatrica, Presidio Infantile Regina Margherita, AOU Città della Salute e della Scienza di Torino
Presidente Associazione Italiana Ematologia e Oncologia Pediatrica (AIEOP)

Le patologie emato-oncologiche in età pediatrica sono malattie rare ma rappresentano la prima causa di morte per malattia e la seconda dopo cause esterne.

Il rischio cumulativo di patologia oncologica fra 0 e 14 anni è di 2,4‰ cioé 1 su 413 nuovi nati si ammaleranno di tumore prima di compiere 15 anni. L'incidenza dei tumori negli adolescenti è maggiore di quella registrata nei bambini di 0-14 anni, in particolare, è del 30% superiore a quella dei bambini al di sotto di 5 anni e quasi il doppio rispetto ai bambini di età 5-9 e 10-14 anni.

I tumori pediatrici più comuni sono le leucemie, i tumori del sistema nervoso centrale e i linfomi. I tumori più frequenti negli adolescenti sono i linfomi di Hodgkin, seguiti da tumori della tiroide, leucemie, tumori delle cellule germinali, linfomi non-Hodgkin, tumori del sistema nervoso centrale, sarcomi delle parti molli e tumori dell'osso. I tumori pediatrici e degli adolescenti presentano caratteristiche biologiche e cliniche diverse dai tumori dell'adulto.

Da molti anni gli oncologi pediatri hanno attivato collaborazioni a livello nazionale e internazionale al fine di condurre studi clinici rilevanti. Queste reti scientifiche e cliniche hanno permesso di ottenere risultati significativi nell'ambito dell'oncologia pediatrica.

A livello nazionale da oltre 40 anni AIEOP (Associazione Italiana Ematologia e Oncologia Pediatrica), riunisce medici, ricercatori e operatori sanitari che si dedicano allo studio e alla cura dei tumori pediatrici. L'obiettivo principale dell'Associazione è il miglioramento dei risultati nel trattamento delle patologie oncoematologiche e della qualità dell'assistenza tramite la promozione della ricerca, l'elaborazione e l'applicazione di protocolli comuni nazionali e internazionali, la divulgazione della conoscenza in questo campo, la formazione degli operatori e la collaborazione con le associazioni dei genitori e di volontariato.

I centri AIEOP sono strutture del SSN dedicate al trattamento delle patologie oncoematologiche in età pediatrica e costituiscono una rete che condivide protocolli di diagnosi e terapia e progetti di ricerca. Le funzioni essenziali dei Centri AIEOP sono la presa in carico multidisciplinare di tutti i pazienti (diagnosi, terapia, fuori terapia) e il coordinamento dell'attività assistenziale globale tramite la collaborazione con il territorio e con le Associazioni di Volontariato. La rete AIEOP è costituita da 53 Centri diffusi su tutto il territorio Nazionale allo scopo di permettere il trattamento del paziente il più possibile vicino al domicilio della famiglia, garantendo l'uniformità del trattamento.

A livello internazionale è stata recentemente istituita una rete di riferimento europea per i tumori pediatrici (EuroPaedCan) coordinata dalla

prof. Ruth Ladenstein (St. Anna Kinderspital & St. Anna Kinderkrebsforschung, Austria) per garantire l'accesso a tutti i pazienti pediatrici con patologia emato-oncologica a Centri altamente specializzati nella gestione e cura di queste patologie.

Questa rete nasce grazie alle precedenti esperienze di progetti europei (ENCCA, PanCare e ExPO-r-Net) e prevede, mediante la collaborazione tra i diversi Centri Europei con specifiche e comprovate competenze, di garantire uniformità di cure e ridurre le disuguaglianze nell'erogazione dei servizi tra i diversi stati europei.

Questa rete include 57 Centri di 17 diverse Nazioni Europee. I Centri Italiani che hanno ottenuto l'endorsement ministeriale sono 9 e rappresentano i principali Centri AIEOP che gestiscono il maggior numero di pazienti pediatrici italiani affetti da patologia emato-oncologica ed alcuni Centri con elevata competenza per specifiche patologie (es. il Centro di Siena per il retinoblastoma).

Centro	
IRCCS Institute Giannina Gaslini – Genoa	Oncoematologia Pediatrica
AOU Meyer – Florence	Oncoematologia Pediatrica
Pediatric Hospital Bambino Gesù, Rome	Oncoematologia Pediatrica/retinoblastoma
MBBM Foundation – pediatrics – S.Gerardo hospital, Monza	Oncoematologia Pediatrica
AO Padua	Oncoematologia Pediatrica
AOU Siena	Oncoematologia Pediatrica/retinoblastoma
AO City of Health and Science – Turin	Oncoematologia Pediatrica
AOU – Perugia	Oncoematologia Pediatrica
Foundation IRCCS Polyclinic San Matteo, Pavia	Oncoematologia Pediatrica

2. La ricognizione dei centri pediatrici italiani in EuroBloodNet

Silverio Perrotta
Dipartimento della Donna, del Bambino e di Chirurgia Generale e Specialistica, Uniuversità degli studi della Campania "L. Vanvitelli", Napoli

EuroBloodNet è il nome con il quale è stata identificata l'ERN delle malattie ematologiche rare (*European Reference Network in Rare Hematological Diseases*). Tale ERN, coordinata dal Prof. Pierre Fenaux dell'Ospedale di Saint Louis di Parigi, è suddivisa in due *hub*:

1) malattie oncologiche della serie mielode e linfoide, di pertinenza dell'adulto e

2) malattie non oncologiche che comprendono le patologie del Globulo Rosso, le insufficienze midollari, le alterazioni della coagulazione ed i disordini ereditari del metabolismo del ferro.

- Ventuno centri Italiani sono entrati a far parte di tale ERN : AOU Università della Campania "L. Vanvitelli, Università Federico II, Università Padova, Pavia, Verona, Siena, Careggi, San Luigi Gonzaga, Policlinico Umberto I, Modena, Bari, Gemelli, Ospedale Maggiore, San Gerardo, Vicenza, Bergamo, Firenze/Pisa, Galliera, Ospedale Cervello, Istituto Gaslini e Rozzano.
- Sette pediatri sono responsabili di tali centri e 9 pediatri sono indicati come vice. Inoltre i pediatri hanno avuto anche la responsabilità di *subnetwork* e di alcuni argomenti trasversali alle varie discipline ematologiche dell'ERN.

	EuroBloodNet Transversal Fields of Action Coordinators	
	Cross border health	
Oncological	Jane Apperley	j.apperley@imperial.ac.uk
Non oncological	Raffaella Colombatti	r.colombatti@gmail.com
ePAG	Amanda Plate	plate@mpeurope.org
	Best practices	
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	Continuing medical education	
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Non oncological	Patricia Aguilar-Martinez	p-martinez@chu-montpellier.fr
ePAG	Jan Geissler	jan@cmiadvocates.net
	Tele-medicine	
Oncological	Andreas Engert	a.engert@uni-koeln.de
Non oncological	Béatrice Gulbis	Beatrice.Gulbis@erasme.ulb.ac.be
ePAG	Sophie Wintrich	swintrich@mds-foundation.org
	Clinical Trials and Research	
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Non oncological	Antonio Piga	antonio.piga@unito.it
ePAG	Angelo Lorio Brunetta	lorisbrunetta@gmail.com

Malattie nefrologiche pediatriche

A cura della Società Italiana di Nefrologia Pediatrica - SINEPE
(Presidente: *Luisa Murer)*

Francesco Emma
Luisa Murer
Carmine Pecoraro

Pubblicazioni

Malattie Nefrologiche Pediatriche

Carmine Pecoraro

Premessa fondamentale per una analisi dettagliata e il *reporting* fedele e il più esaustivo possibile della ricerca sulle malattie renali rare che viene svolta in Italia nel bambino, dai principali centri di Nefrologia Pediatrica, e della relativa produzione scientifica, è chiarire la differenza di identificazione di malattia renale rara come definita nell' "Elenco delle Malattie Rare esente da Ticket" pubblicato dal Centro Nazionale Malattie Rare dell'ISS (www.iss.it/cnmr) in base al DM 279/2001 e le categorie individuate dalla European Renal Network per le malattie renali rare (ERKnet). Nel datato elenco le malattie sono indicate, una ad una, con un codice di raggruppamento di organo e/o apparato che, alla luce delle nuove conoscenze, sono, non di rado, collocate nel gruppo sbagliato oltre al fatto di essere sottorappresentate in misura significativa.

Anche gli aggiornamenti più recenti effettuati dalle regioni, come quello fatto dalla regione Campania, sono insufficienti a coprire l'intero spettro delle malattie renali del bambino. Diversi e assolutamente più rappresentativi della ampia ed eterogenea gamma delle malattie renali sono stati i criteri che hanno informato la indagine tra i paesi europei che il coordinatore della ERKnet, Franz Schaefer di Heidelberg, ha condotto per individuare i centri che soddisfacessero i requisiti minimi di qualità dell'assistenza e della ricerca da ammettere nella rete stessa. Sono stati individuate 10 categorie di malattia, es. Hereditary Glomerulopathies, Immunological Glomerulopathies, Tubulopathies, etc, includendo anche condizioni come Pediatric kidney TX e Pediatric Chronic Kidney Disease 3-5 and Dialysis. Sono stati ammessi, alla fine delle procedure, 39 Health Care Providers, tra centri esclusivamente padiatrici e centri adulti, con diverso numero di categorie di malattia. Orbene, il report dello stato dell'arte della produzione scientifica più recente e sullo sviluppo dei farmaci orfani per le malattie renali rare con la partecipazione alle sperimentazioni di gruppi italiani, verrà condotto ispirandosi alle categorie comprese nella ERKnet.

Pubblicazioni

BERGAMO, Mario Negri

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
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	Mele C, Lemaire M, Iatropoulos P, Piras R, Bresin E, Bettoni S, Bick D, Helbling D, Veith R, Valoti E, Donadelli R, Murer L, Neunhäuserer M, Breno M, Frémeaux-Bacchi V, Lifton R, Remuzzi G, *Noris M*.Clin	Characterization of a New DGKE Intronic Mutation in Genetically Unsolved Cases of Familial Atypical Hemolytic Uremic Syndrome.	J Am Soc Nephrol.	2015		
	Nester CM, Barbour T, de Cordoba SR, Dragon-Durey MA, Fremeaux-Bacchi V, Goodship TH, Kavanagh D, *Noris M*, Pickering M, Sanchez-Corral P, Skerka C, Zipfel P, Smith RJ.	Atypical aHUS: State of the art.	Mol Immunol.	2015		
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	Coppo R, Peruzzi L, Amore A, Martino S, Vergano L, Lastauka I, Schieppati A, *Noris M*, Tovo PA, Remuzzi G.	Dramatic effects of eculizumab in a child with diffuse proliferative lupus nephritis resistant to conventional therapy.	Pediatr Nephrol.	2015		

	Noris M*, Galbusera M, Gastoldi S, Macor P, Banterla F, Bresin E, Tripodo C, Bettoni S, Donadelli R, Valoti E, Tedesco F, Amore A, Coppo R, Ruggenenti P, Gotti E, Remuzzi G.	Dynamics of complement activation in aHUS and how to monitor eculizumab therapy	Blood.	2014		
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OSPEDALE PEDIATRICO BAMBINO GESU' - ROMA

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Cystinosis	Taranta A, Bellomo F, Petrini S, Polishchuk E, De Leo E, Rega LR, Pastore A, Polishchuk R, De Matteis MA, Emma F	Cystinosin-LKG Rescues Cystine Accumulation and Decreases Apoptosis Rate in Cystinotic Proximal Tubular Epithelial Cells	Pediatr Res. 81(1-1):113-119.	2017	2,76	
	Data From the European Society for Pediatric Nephrology/European Renal Association-European Dialysis and Transplant (ESPN/ERA-EDTA) Registry. Mekahli D, van Stralen KJ, Bonthuis M, Jager KJ, Balat A, Benetti E, Godefroid N, Edvardsson VO, Heaf	Kidney Versus Combined Kidney and Liver Transplantation in Young People With Autosomal Recessive Polycystic Kidney Disease:	Am J Kidney Dis.	2016		

	JG, Jankauskiene A, Kerecuk L, Marinova S, Puteo F, Seeman T, Zurowska A, Pirenne J, Schaefer F, Groothoff JW; ESPN/ERA-EDTA Registry					
Cystinosis	Bellomo F, Taranta A, Petrini S, Venditti R, Rocchetti MT, Rega LR, Corallini S, Gesualdo L, De Matteis MA, Emma F	Carboxyl-Terminal SSLKG Motif of the Human Cystinosin-LKG Plays an Important Role in Plasma Membrane Sorting	PLoS One. 11(5):e0154805.	2016	3,06	
Cystinosis	Rega LR, Polishchuk E, Montefusco S, Napolitano G, Tozzi G, Zhang J, Bellomo F, Taranta A, Pastore A, Polishchuk R, Piemonte F, Medina DL, Catz SD, Ballabio A, Emma F	Activation of the transcription factor EB rescues lysosomal abnormalities in cystinotic kidney cells	Kidney Int. 89(4):862-73.	2016	7,68	
Idiopathic Nephrotic Syndrome	Colucci M, Carsetti R, Cascioli S, Casiraghi F, Perna A, Ravà L, Ruggiero B, Emma F, Vivarelli M	B Cell Reconstitution after Rituximab Treatment in Idiopathic Nephrotic Syndrome	J Am Soc Nephrol. 27(6):1811-22.	2016	9,34	
Renal disease, general	Emma F, Montini G, Parikh SM, Salviati L	Mitochondrial dysfunction in inherited renal disease and acute kidney injury	Nat Rev Nephrol. 12(5):267-80.	2016	9,46	
	Vivarelli M, Massella L, Ruggiero B, Emma F.	Minimal Change Disease.	Clin J Am Soc Nephrol.	2016		
	Dolcemascolo V, Vivarelli M, Colucci M, Diomedi-Camassei F, Piras R, Alberti M, Emma F.	Nephrotic-Range Proteinuria and Peripheral Edema in a Child: Not Only Idiopathic Nephrotic Syndrome	Case Rep Nephrol Dial.	2016		
	Bacchetta J, Greco M, Bertholet-Thomas A, Nobili F, Zustin J, Cochat P, Emma F, Boivin G. Bonekey	Skeletal implications and management of cystinosis: three case reports and literature review.	Rep. 2016	2016		

	an Coppo R, Lofaro D, Camilla RR, Bellur S, Cattran D, Cook HT, Roberts IS, Peruzzi L, Amore A, Emma F, Fuiano L, Berg U, Topaloglu R, Bilginer Y, Gesualdo L, Polci R, Mizerska-Wasiak M, Caliskan Y, Lundberg S, Cancarini G, Geddes C, Wetzels J, Wiecek A, Durlik M, Cusinato S, Rollino C, Maggio M, Praga M, K Smerud H, Tesar V, Maixnerova D, Barratt J, Papalia T, Bonofiglio R, Mazzucco G, Giannakakis C, Soderberg M, Orhan D, Di Palma AM, Maldyk J, Ozluk Y, Sudelin B, Tardanico R, Kipgen D, Steenbergen E, Karkoszka H, Perkowska-Ptasinska A, Ferrario F, Gutierrez E, Honsova E.	7. Risk factors for progression in children and young adults with IgA nephropathy: analysis of 261 cases from the VALIGA European cohort	Pediatr Nephrol. 2017 Jan;32(1):139-150. Erratum in: Pediatr Nephrol	2016		
	Ruggiero B, Vivarelli M, Gianviti A, Pecoraro C, Peruzzi L, Benetti E, Ventura G, Pennesi M, Murer L, Coppo R, Emma F	Outcome of childhood-onset full-house nephropathy.	Nephrol Dial Transplant.	2016		
	. Langman CB, Barshop BA, Deschênes G, Emma F, Goodyer P, Lipkin G, Midgley JP, Ottolenghi C, Servais A, Soliman NA, Thoene JG, Levtchenko EN; Conference Participants.	Controversies and research agenda in nephropathic cystinosis: conclusions from a "Kidney Disease: Improving Global Outcomes" (KDIGO) Controversies Conference	Kidney Int. 2016 Jun	2016		

	Bellomo F, Taranta A, Petrini S, Venditti R, Rocchetti MT, Rega LR, Corallini S, Gesualdo L, De Matteis MA, Emma F	Carboxyl-Terminal SSLKG Motif of the Human Cystinosin-LKG Plays an Important Role in Plasma Membrane Sorting.	PLoS One. 2016 May	2016		
	Rega LR, Polishchuk E, Montefusco S, Napolitano G, Tozzi G, Zhang J, Bellomo F, Taranta A, Pastore A, Polishchuk R, Piemonte F, Medina DL, Catz SD, Ballabio A, Emma F	Activation of the transcription factor EB rescues lysosomal abnormalities in cystinotic kidney cells.	Kidney Int. 2016	2016		
	Gansevoort RT, Arici M, Benzing T, Birn H, Capasso G, Covic A, Devuyst O, Drechsler C, Eckardt KU, Emma F, Knebelmann B, Le Meur Y, Massy ZA, Ong AC, Ortiz A, Schaefer F, Torra R, Vanholder R, Więcek A, Zoccali C, Van Biesen W.	Recommendations for the use of tolvaptan in autosomal dominant polycystic kidney disease: a position statement on behalf of the ERA-EDTA Working Groups on Inherited Kidney Disorders and European Renal Best Practice.	Nephrol Dial Transplant	2016		
	Iatropoulos P, Noris M, Mele C, Piras R, Valoti E, Bresin E, Curreri M, Mondo E, Zito A, Gamba S, Bettoni S, Murer L, Fremeaux-Bacchi V, Vivarelli M, Emma F, Daina E, Remuzzi G	Complement gene variants determine the risk of immunoglobulin-associated MPGN and C3 glomerulopathy and predict long-term renal outcome	Mol Immunol. 2016	2016		
	Emma F, Montini G, Parikh SM, Salviati L.	Mitochondrial dysfunction in inherited renal disease and acute kidney injury	Nat Rev Nephrol. 2016	2016		
	Barbour SJ, Espino-Hernandez G, Reich HN, Coppo R, Roberts IS, Feehally J, Herzenberg AM, Cattran DC; Oxford Derivation, North American Validation and VALIGA Consortia.; Oxford Derivation North American Validation and VALIGA Consortia	The MEST score provides earlier risk prediction in lgA nephropathy.	Kidney Int.	2016		

	Santilli V, Cagigi A, Guzzo I, Rinaldi S, Mora N, Zotta F, Piazza A, Rossi P, Emma F, Dello Strologo L, Palma P.	Cellular immune profile of kidney transplant patients developing anti-HLA antibodies during childhood.	Pediatr Nephrol.	2016		
	Chinali M, Emma F, Esposito C, Rinelli G, Franceschini A, Doyon A, Raimondi F, Pongiglione G, Schaefer F, Matteucci MC	Left Ventricular Mass Indexing in Infants, Children, and Adolescents: A Simplified Approach for the Identification of Left Ventricular Hypertrophy in Clinical Practice.	J Pediatr.	2016		
	Feehally J, Coppo R, Troyanov S, Bellur SS, Cattran D, Cook T, Roberts IS, Verhave JC, Camilla R, Vergano L, Egido J, Wiecek A, Karkoszka H, Tesar V, Maixnerova D, Ots-Rosenberg M, Quaglia M, Rollino C, Magistroni R, Cusinato S, Cravero R, Peruzzi L, Lundberg S, Gesualdo L, Cancarini G, Feriozzi S, Ferrario F	Tonsillectomy in a European Cohort of 1,147 Patients with IgA Nephropathy.	VALIGA study of ERA-EDTA Immunonephrology Working Group. Nephron	2016		
	Colucci M, Carsetti R, Cascioli S, Casiraghi F, Perna A, Ravà L, Ruggiero B, Emma F, Vivarelli M	B Cell Reconstitution after Rituximab Treatment in Idiopathic Nephrotic Syndrome	J Am Soc Nephrol.	2016		
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Chronic kidney disease	Chinali M, Matteucci M, Franceschini A, Doyon A, Pongiglione G, Rinelli G, Schaefer F	Advanced parameters of cardiac mechanics in children with CKD: The 4C study	Clin J Am Soc Nephrol. 10(8):1357-63.	2015	4,61	
Cystinosis	Conforti A, Taranta A, Biagini S, Starc N, Pitisci A, Bellomo F, Cirillo V, Locatelli F, Bernardo M, Emma F	Cysteamine treatment restores the in vitro ability to differentiate along the osteoblastic lineage of mesenchymal stromal cells isolated from bone marrow of a cystinotic patient	J Transl Med. 13:143.	2015	3,93	
Nephropathy	Vivarelli M, Emma F, Pellé T, Gerken C, Pedicelli S, Diomedi-Camassei F, Klaus G, Waldegger S, Ronco P, Debiec H	Genetic homogeneity but IgG subclass-dependent clinical variability of alloimmune membranous nephropathy with anti-neutral endopeptidase antibodies	Kidney Int. 87(3):602-9.	2015	8,56	

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Chronic granulomatosis	Claps A, Della Corte M, Gerocarni Nappo S, Francalanci P, Palma P, Finocchi A	How should eosinophilic cystitis be treated in patients with chronic granulomatous disease?	Pediatr Nephrol. 9(11):2229-33.	2014	0	see IMMUNOLOGICAL

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Cystinosis	Prencipe G, Caiello I, Cherqui S, Whisenant T, Petrini S, Emma F, De Benedetti F	Inflammasome activation by cystine crystals: implications for the pathogenesis of cystinosis	J Am Soc Nephrol. 25(6):1163-9.	2014	9,47	see METABOLIC
Glomerulopathy	Vivarelli M and Emma F	Treatment of C3 glomerulopathy with complement blockers	Semin Thromb Hemost. 40(4):472-7.	2014	3,69	
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	Spithoven EM, Kramer A, Meijer E, Orskov B, Wanner C, Abad JM, Aresté N, de la Torre RA, Caskey F, Couchoud C, Finne P, Heaf J, Hoitsma A, de Meester J, Pascual J, Postorino M, Ravani P, Zurriaga O, Jager KJ, Gansevoort RT; ERA-EDTA Registry; EuroCYST Consortium; WGIKD	Renal replacement therapy for autosomal dominant polycystic kidney disease (ADPKD) in Europe: prevalence and survival--an analysis of data from the ERA-EDTA Registry	Nephrol Dial Transplant	2014		

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	Maiorana A, Vergine G, Coletti V, Luciani M, Rizzo C, Emma F, Dionisi-Vici C. Nutrition	Acute thiamine deficiency and refeeding syndrome: Similar findings but different pathogenesis.	Nutrition	2014		
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	Spithoven EM, Kramer A, Meijer E, Orskov B, Wanner C, Caskey F, Collart F, Finne P, Fogarty DG, Groothoff JW, Hoitsma A, Nogier MB, Postorino M, Ravani P, Zurriaga O, Jager KJ, Gansevoort RT	Analysis of data from the ERA-EDTA Registry indicates that conventional treatments for chronic kidney disease do not reduce the need for renal replacement therapy in autosomal dominant polycystic kidney disease	Kidney Int.	2014		

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Idiopathic nephrotic syndrome	Ruggenenti P, Ruggiero B, Cravedi P, Vivarelli M, Massella L, Marasà M, Chianca A, Rubis N, Ene-Iordache B, Rudnicki M, Pollastro R, Capasso G, Pisani A, Pennesi M, Emma F, Remuzzi G	Rituximab in steroid-dependent or frequently relapsing idiopathic nephrotic syndrome.	J Am Soc Nephrol. 25(4):850-63.	2014	9,47	
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	Ferraresso M, Belingheri M, Ginevri F, Murer L, Dello Strologo L, Cardillo M, Parodi A, Ghirardo G, Guzzo I, Innocente A, Ghio L	Three-yr safety and efficacy of everolimus and low-dose cyclosporine in de novo pediatric kidney transplant patients	Pediatr Transplant	2014		
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	Harambat J, van Stralen KJ, Verrina E, Groothoff JW, Schaefer F, Jager KJ	Likelihood of children with end-stage kidney disease in Europe to live with a functioning kidney transplant is mainly explained by nonmedical factors.	Pediatr Nephrol	2014		
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Ospedale Padova

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STRUTTURA COMPLESSA DI NEFROLOGIA E DIALISI A.O SANTOBONO-PAUSILIPON NAPOLI

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	Stefano Picca, Carlo Dionisi-Vici, Andrea Bartuli ,Tommaso De Palo ,Francesco Papadia ,Giovanni Montini, Marco Materassi, Maria Alice Donati, Enrico Verrina, Maria Cristina Schiaffino,Carmine Pecoraro, Emilia Iaccarino, Enrico Vidal, Alberto Burlina, Francesco Emma	Short-term survival of hyperammonemic neonates treated with dialysis	Pediatric Nephrology	2014		
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	Carmine Pecoraro Alfonso Vincenzo Salvatore Ferretti Erica Rurali • [...] Giuseppe Remuzzi	Treatment of congenital thrombotic thrombocytopenic purpura with eculizumab	Pediatric Nephrology	2015		
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	Carmine Pecoraro	Prevention of chronic kidney disease (ckd) in children	Italian Journal of Pediatrics	2015		
	Chantal Loirat ,Fadi Fakhouri ,Gema Ariceta ,Carmine Pecoraro [...] , Véronique Frémeaux-Bacchi	An international consensus approach to the management of atypical hemolytic uremic syndrome in children	Pediatric Nephrology	2015		
	G. Malgieri · V. Bruno · A. Esposito C. Pecoraro ·	Nutcracker syndrome: an underestimated cause of hematuria in children	Pediatric Nephrology	2016		
	V. Bruno ,D. Molino ,F. Nuzzi , [...] C. Pecoraro	Mycophenolate Mofetil (MMF) as induction and maintenance therapy in childhood Lupus Nephritis (LN)	Pediatric Nephrology	2016		

	E. Vidal ,I. Alberici ,M. Martino, C.Pecoraro [...], E. Verrina	Acute pancreatitis in children on chronic dialysis: incidence and clinical characteristic in a nationwide registry.	Pediatric Nephrology	2016		
	L. Peruzzi , B. Gianoglio , C. Pecoraro, [...] , R. Coppo	EUROPEAN REGISTRY OF HENOCH SCHOENLEINPURPURA NEPHRITIS IN CHILDREN TO DETECT RISK…	Nephrology Dialysis	2016		
	Barbara Ruggiero ,Marina Vivarelli , Alessandra Gianviti ,C.Pecoraro [...] ,Francesco Emma	Outcome of childhood-onset full-house nephropathy	Nephrology Dialysis Transplantation	2016		
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Ospedale Regina Margherita - Torino

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IgA nephropathy	Coppo R, Lofaro DD, Camilla RR, Bellur S, Cattran D, Cook HT, Roberts IS, Peruzzi L, Amore A, Emma F, Fuiano L, Berg U, Topaloglu R, Bilginer Y, Gesualdo L, Polci R, Mizerska-Wasiak M, Caliskan Y, Lundberg S, Cancarini G, Geddes C, Wetzels J, Wiecek A, Durlik M, Cusinato S, Rollino C, Maggio M, Praga MK, Smerud H, Tesar V, Maixnerova D, Barratt J, Papalia T, Bonofiglio R ,Mazzucco	Risk factors for progression in children and young adults with IgA nephropathy: an analysis of 261 cases from the VALIGA European cohort	Pediatr Nephrol. 32(1):139-150.	2017	2,34	

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Partecipazione Nazionale a ERN

Francesco Emma
Divisione di Nefrologia e Dialisi Ospedale Pediatrico Bambino Gesù

La rete ERN per le malattie renali si nomina ERKNeT ed è coordinata dal Prof. F. Schaefer presso l'università di Heidelberg.
In base ai dati di prevalenza disponibili nei registri adulti e pediatrici delle rispettive società europee di nefrologia, le malattie sono state classificate in nove sottocategorie che comprendono:

1. Glomerulopatie
2. Malattie congenite dei reni e delle vie urinarie e ciliopatie
3. Displasie renali autosomiche dominanti e patologie correlate
4. Tubulopathie
5. Malattie metaboliche e disordini caratterizzati da nefrolitiasi
6. Microangiopatie trombotiche
7. Insufficienza renale cronica di stadio 3-5 in età pediatrica e dialisi pediatrica
8. Trapianto renale pediatrico

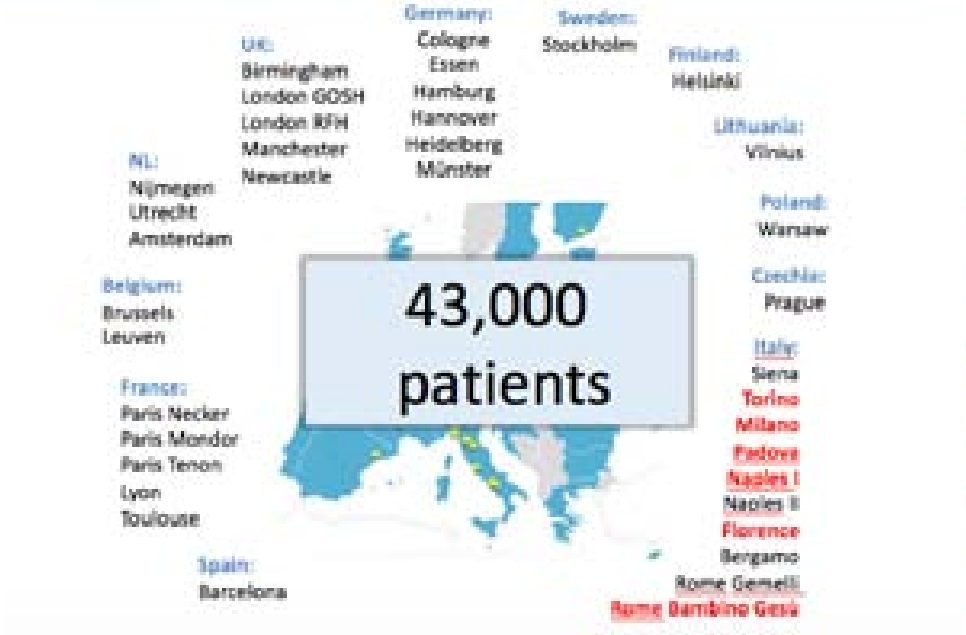
Per aderire alla rete, i centri hanno dovuto certificare di seguire un minimo di pazienti in una o più categoria o di avere una produzione scientifica e/o un ruolo di leader in registri o studi clinici inerenti alla specifica categoria.

È stato dunque creato un punteggio che ha permesso ai vari centri di qualificarsi come centro di riferimento in almeno una delle nove categorie identificate. In Italia, i centri pediatrici che afferiscono al momento alla rete ERKNeT sono: Torino (Ospedale Regina Margherita), Milano (Fondazione IRCCS Ca' Granda - Ospedale Maggiore Policlinico), Padova (Clinica Pediatrica – Azienda Ospedaliera di Padova), Firenze (Ospedale Meyer), Roma (Ospedale Pediatrico Bambino Gesù) e Napoli (Ospedale Santo Bono).
Alle attività delle varie aree tematiche, contribuiscono 6 task force trasversali per:

1. Diagnostica molecolare
2. Registri e valutazioni di esito
3. Anatomia patologica ed imaging
4. Transizione bambino-adulto e qualità di vita
5. Formazione e training
6. Linee guida

Il mantenimento di ogni centro all'interno della rete è condizionato alla partecipazione attiva alle suddette attività ed al raggiungimento di criteri di benchmarking elaborati dal consorzio.

ERKNet European Rare Kidney Disease Reference Network



ERKNet: Scope of Diseases

Ped > Adult
Ped = Adult
Ped < Adult

Glomerulopathies
Idiopathic NS
SRNS/FSGS
Alport
Fabry
MPGN
Membranous GN
Other immune-mediated GN

CAKUT & Ciliopathies
Syndromic
Renal hypo/dysplasia
ARPKD
Nephronophthisis
Bardet Biedl Syndrome
PUV/obstructive uropathies

AD renal dysplasia disorders
ADPKD
Tuberous sclerosis, VHL
ADTKD (UMOD, HNF1B, Muc1, Ren)

Tubulopathies
Renal tubular acidosis
Bartter-like disorders
Disorders of Mg homeostasis
Disorders of Ca homeostasis
Disorders of P homeostasis
Dent's disease
Lowe syndrome

Metabolic and stone forming disorders
Primary hyperoxaluria
Cystinosis
Cystinuria
MMA

Thrombotic microangiopathies

Pediatric CKD stage 3-5 and Dialysis

Malattie immunologiche pediatriche

A cura del Gruppo di Immunodeficienze - IPINET-AIEOP
*(*Coordinatore: *Claudio Pignata)*

Alessandro Aiuti
Marco Gattorno
Claudio Pignata
Alessandro Plebani

Pubblicazioni, *trial* clinici e ERN

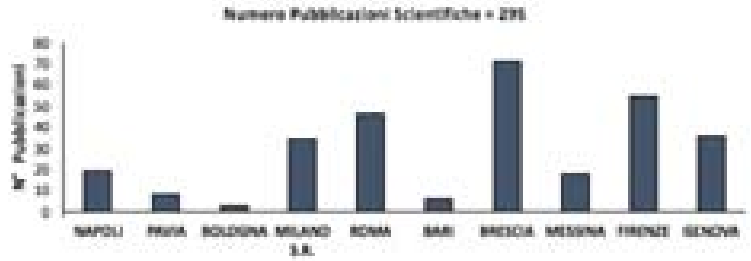
Claudio Pignata, Alessandro Aiuti, Alessandro Plebani

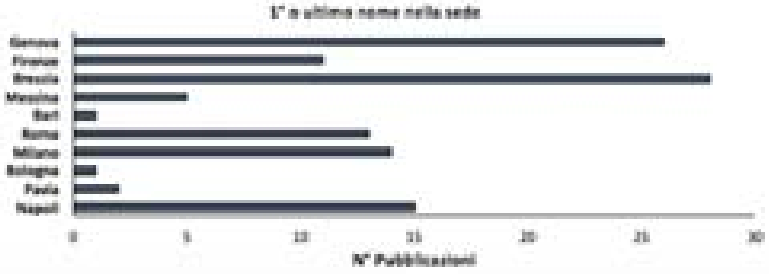
I disordini congeniti del sistema immune rappresentano una frontiera della nuova medicina traslazionale e della ricerca in ambito biomedico. La maggior parte delle scoperte che hanno riguardato negli ultimi anni questo settore della pediatria ha aperto la strada per una migliore comprensione dei meccanismi fisiopatologici, biochimici e molecolari, di molti disordini rari del sistema immune. Ciò è stato reso possibile dalla creazione di un *Network* di Centri italiani dedicati a questo gruppo di malattie rare. La preparazione di protocolli di studio retro-prospettici (al momento 11) ha permesso di ottenere informazioni sulla storia naturale in coorti di pazienti affetti da malattie molto rare, ponendo il nostro Paese all'avanguardia in questo ambito. Il riconoscimento di specifici meccanismi patogenetici ha consentito lo sviluppo di approcci terapeutici innovativi personalizzati basati sul meccanismo di malattia del singolo paziente, consentendo a molti di essi, di essere curati in maniera definitiva.

Al fine di valutare i risultati dei vari gruppi che in Italia si occupano della gestione diagnostico terapeutica e di ricerca in tale settore, è stata effettuata una valutazione della produzione scientifica dell'ultimo triennio mediante l'utilizzo del motore di ricerca PubMed**,** basato sul database MEDLINE. Per ogni produzione scientifica è stato calcolato l'indice scientometrico, quale l'Impact Factor, che ha permesso di acquisire informazioni sul numero di pubblicazioni su riviste con IF > 4 < 10 e > 10, individuando anche i gruppi che hanno capacità autoctone, come dimostrato dalla presenza di primo o ultimo autore della pubblicazione. Inoltre, mediante l'indice di Hirsch (H-index), è stato possibile quantizzare l'impatto scientifico di ciascun autore. Tale analisi ha consentito altresì di valutare la presenza dei principali ricercatori italiani nel settore tra i Top Italian Scientists (TIS) e nei database di *ranking* professionale internazionali (Expertscape).

Nel *contest* della partecipazione alla ERN RITA (che include anche le malattie autoinfiammatorie, in Italia assistite dai Centri di Reumatologia), sono stati identificati i Centri che partecipano come *Core Centers* e i Centri che in questa prima fase sono *Affiliated*. È stata infine condotta un'analisi dei Centri che, avendo ottenuto l'*endorsement* ministeriale quali centri delle immunodeficienze, consentono di formalizzare la mappatura nazionale dei Centri con *expertise*.

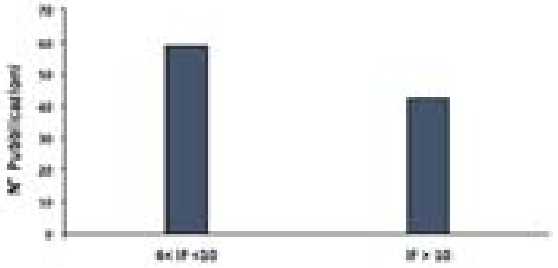
Valutazione scientifica nell'ultimo triennio





Valutazione scientifica nell'ultimo triennio

Impact Factor Totale = 1761



Pubblicazioni

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
APDS	Chiriaco M, Brigida I, Ariganello P, Di Cesare S, Di Matteo G, Taus F, Cittaro D, Lazarevic D, Scarselli A, Santilli V, Attardi E, Stupka E, Giannelli S, Fraziano M, Finocchi A, Rossi P, Aiuti A, Palma P, **Cancrini C**	A case of APDS patient: defects in maturation and function and decreased in vitro anti-mycobacterial activity in the myeloid compartment	Clin Immunol.	2015	4,03	OPBG
Agammaglobulinemia	Cifaldi C, Scarselli A, Petricone D, Di Cesare S, Chiriaco M, Claps A, Rossi P, Calzoni E, Yamazaki Y, Notarangelo LD, Di Matteo G, Cancrini C, **Finocchi A**	Agammaglobulinemia associated to nasal polyposis due to a hypomorphic RAG1 mutation in a 12 years old boy	Clin Immunol. pii: S1521 6616(16)30410-7	2016	4,03	OPBG
Immunological disorder, general	Aranburu A, Piano Mortari E, Baban A, Giorda E, Cascioli S, Marcellini V, Scarsella M, Ceccarelli S, Corbelli S, Cantarutti N, De Vito R, Inserra A, Nicolosi L, Lanfranchi A, Porta F, Cancrini C, Finocchi A, **Carsetti R**	Human B-cell memory is shaped by age- and tissue-specific T-independent and GC-dependent events	Eur J Immunol. 2016 Nov 16	2016	4,18	OPBG
PID	**Cifaldi L,** Pinto RM, Rana I, Caniglia M, Angioni A, Petrocchi S, Cancrini C, Cursi L, Palumbo G, Zingoni A, Gismondi A, Rossi P, Santoni A, Cerboni C	NK cell effector functions in a Chédiak-Higashi patient undergoing cord blood transplantation: Effects of in vitro treatment with IL-2	Immunol Lett. 2016 Dec;180:46-53	2016	2,483	OPBG

Ataxia-Teleangiectasia	Cantarutti N, Claps A, Angelino G, Chessa L, Callea F, El Hachem M, Diociaiuti A, **Finocchi A**	Multi-drugs resistant acne rosacea in a child affected by Ataxia-Telangiectasia: successful treatment with Isotretinoin	Ital J Pediatr.41:23. doi: 10.1186/s13052-015-0125-7	2015	1,614	OPBG
CGD	**Cotugno N,** Finocchi A, Cagigi A, Di Matteo G, Chiriaco M, Di Cesare S, Rossi P, Aiuti A, Palma P, Douagi I	Defective B-cell proliferation and maintenance of long-term memory in patients with chronic granulomatous disease	J Allergy Clin Immunol. 135(3):753-61.e2	2015	11,48	OPBG
22q11.2 deletion syndrome	Di Cesare S, Puliafito P, Ariganello P, Marcovecchio GE, Mandolesi M, Capolino R, Digilio MC, Aiuti A, Rossi P, **Cancrini C**	Autoimmunity and regulatory T cells in 22q11.2 deletion syndrome patients	Pediatr Allergy Immunol. 226(6):591-4	2015	3,4	OPBG
Autoimmunity and immunodeficiency	**Bellacchio E**, Palma A, Corrente S, Di Girolamo F, Helen Kemp E, Di Matteo G, Comelli L, Carsetti R, Cascioli S, Cancrini C, **Fierabracci A**	The possible implication of the S250C variant of the autoimmune regulator protein in a patient with autoimmunity and immunodeficiency: in silico analysis suggests a molecular pathogenic mechanism for the variant	Gene. 2549(2):286-94	2014	2,415	OPBG
Antibody deficiency	Di Pierro V, Zuntini R, Cancrini C, Finocchi A, Angelino G, Rossi P, **Ferrari S**	Consanguinity and polygenic diseases: a model for antibody deficiencies	Hum Hered. ;77(1-4):144-9	2014		S.Orsola Malpighi Bologna

22q11.2 deletion syndrome	**Cancrini C**, Puliafito P, Digilio MC, Soresina A, Martino S, Rondelli R, Consolini R, Ruga EM, Cardinale F, Finocchi A, Romiti ML, Martire B, Bacchetta R, Albano V, Carotti A, Specchia F, Montin D, Cirillo E, Cocchi G, Trizzino A, Bossi G, Milanesi O, Azzari C, Corsello G, Pignata C, Aiuti A, Pietrogrande MC, Marino B, Ugazio AG, Plebani A, Rossi P; Italian Network for Primary Immunodeficiencies	Clinical features and follow-up in patients with 22q11.2 deletion syndrome	J Pediatr.164(6):1475-80.e2	2014	3,89	OPBG
Neutropenia	Angelino G, Caruso R, D'Argenio P, Calò Carducci FI, Pascone R, Lanciotti M, Cancrini C, Palma P, Aiuti A, Rossi P, **Finocchi A**	Etiology, clinical outcome, and laboratory features in children with neutropenia: analysis of 104 cases	Pediatr Allergy Immunol. 25(3):283-9	2014	3,4	OPBG
CGD	Chiriaco M, Salfa I, Matteo GD, Rossi P, **Finocchi A**	Chronic Granulomatous Disease: clinical, molecular and therapeutic aspects	Pediatr Allergy Immunol. 27(3):242-53	2016	3,40	OPBG
CGD	Claps A, Della Corte M, Gerocarni Nappo S, Francalanci P, Palma P, **Finocchi A**	How should eosinophilic cystitis be treated in patients with chronic granulomatous disease?	Pediatr Nephrol. 9(11):2229-33	2014	2,516	OPBG
CGD	**Finocchi A**, Claps A, Serafinelli J, Salfa I, Longo D, Di Matteo G, Aiuti A, Rossi P	Chronic granulomatous disease presenting with salmonella brain abscesses	Pediatr Infect Dis J. 33(5):525-8	2014	3,14	OPBG

CID	Cifaldi C ,Angelino G, Chiriaco M, Di Cesare S, Claps A, Serafinelli J, Rossi P, Antoccia A, Di Matteo G, Cancrini C, De Villartay JP, **Finocchi A**	Late-onset combined immune-deficiency due to LIGIV mutations in a 12 years old patient	Pediatr Allergy Immunol. 28(2):203-206	2017	3,95	OPBG
PID	Marasco E, Farroni C, Cascioli S, Marcellini V, Scarsella M, Giorda E, Piano Mortari E, Leonardi L, Scarselli A, Valentini D, Cancrini C, Duse M, Grimsholm O, **Carsetti R**	B-cell activation with CD40L or CpG measures the function of B-cell subsets and identifies specific defects in immunodeficient patients	Eur J Immunol. 47(1):131-143	2017	4,18	OPBG
CID	Scarselli A, Di Cesare S, Di Matteo G, De Matteis A, Ariganello P, Romiti ML, Cascioli S, De Vito R, Bertaina A, Locatelli F, Gaspar HB, Aiuti A, Rossi P, Gilmour K, **Cancrini C**	Combined immunodeficiency due to JAK3 mutation in a child presenting with skin granuloma	J Allergy Clin Immunol. 137(3):948-51.e5	2016	11,48	OPBG
PID	Vultaggio A, Azzari C, Milito C, Finocchi A, Toppino C, Spadaro G, Trizzino A, Baldassarre M, Paganelli R, Moschese V, Soresina A, Matucci A	Subcutaneous Immunoglobulin Replacement Therapy in Patients with Primary Immunodeficiency in Routine Clinical Practice: The VISPO Prospective Multicenter Study	Clin Drug Investig. 2015 Mar;35(3):179-85	2015	1,56	Careggi Firenze
Immunological disorder, general	Brigida I, Chiriaco M, Di Cesare S, Cittaro D, Di Matteo G, Giannelli S, Lazarevic D, Zoccolillo M, Stupka E, Jenkner A, Francalanci P, Livadiotti S, Morawski A, Ravell J, Lenardo M, Cancrini C, Aiuti A, **Finocchi A**	Large Deletion of MAGT1 Gene in a Patient with Classic Kaposi Sarcoma, CD4 Lymphopenia, and EBV Infection	J Clin Immunol. 37(1):32-35.	2017	3,09	OPBG

Autoimmunity and immunodeficiency	Zama D, Cocchi I, Masetti R, Specchia F, Alvisi P, Gambineri E, Lima M, **Pession A**	Late-onset of immunodysregulation, polyendocrinopathy, enteropathy, x-linked syndrome (IPEX) with intractable diarrhea.	Ital J Pediatr	2014	1,614	Sant'Orsola Malpighi,Bologna
PID	Pulvirenti F, Zuntini R, Milito C, Specchia F, Spadaro G, Danieli MG, Pession A, Quinti I, **Ferrari S**	Clinical Associations of Biallelic and Monoallelic TNFRSF13B Variants in Italian Primary Antibody Deficiency Syndromes.	J Immunol Res		3,276	S.Orsola Malpighi Bologna
Agammaglobulinemia	**Dellepiane R.M.**, Dell'Era L., Beilis L.V., Pavesi P., Raimondi M., Soresina A., Lougaris V., Carrabba M.,Martire B., Martino S., Russo G., Patuzzo G., Pignata C., Spadaro G., Gallizzi R., Duse M., Specchia F.G.,Moschese V., Marseglia G.L., Pietrogrande M.C., Bedogni G., Agostoni C	Nutritional Status in Agammaglobulinemia: An Italian Multicenter Study	JClin Immunol. 35(7):595-7	2015	3.094	Policlinico Ca Granda Milano
Complement deficiency	**Dellepiane R.M.**, Dell'Era L., Pavesi P., Macor P., Giordano M., De Maso L., Pietrogrande M.C., Cugno M	Invasive meningococcal disease in three siblings with hereditary deficiency of the 8(th) component of complement: evidence for the importance of an early diagnosis	Orphanet J Rare Dis. 11(1):64-69	2016	3.290	Policlinico Ca Granda Milano

ADA-SCID	Carriglio N, Klapwijk J, Hernandez RJ, Vezzoli M, Chanut F, Lowe R, Elena D, Nord M, Albertini P, Cristofori P, Richards J, Staton H, Appleby J, **Aiuti A**, Sauer AV.	Good Laboratory Practice Preclinical Safety Studies for GSK2696273 (MLV Vector-Based Ex Vivo Gene Therapy for Adenosine Deaminase Deficiency Severe Combined Immunodeficiency) in NSG Mice.	Hum Gene Ther Clin Dev 28:17-27	2017	2,889	San Raffaele-Milano
ADA-SCID	Sauer AV, Hernandez RJ, Fumagalli F, Bianchi V, Poliani PL, Dallatomasina C, Riboni E, Politi LS, Tabucchi A, Carlucci F, Casiraghi M, Carriglio N, Cominelli M, Forcellini CA, Barzaghi F, Ferrua F, Minicucci F, Medaglini S, Leocani L, la Marca G, Notarangelo LD, Azzari C, Comi G, Baldoli C, Canale S, Sessa M, D'Adamo P, **Aiuti A.**	Alterations in the brain adenosine metabolism cause behavioral and neurological impairment in ADA-deficient mice and patients	Sci Rep., 7: 40136	2017	5,228	San Raffaele-Milano
PID	Ingo DM, Redaelli D, Rossella V, Perini O, Santoleri L, Ciceri F, **Aiuti A, Bernardo ME**	Bone marrow-derived CD34-fraction: A rich source of mesenchymal stromal cells for clinical application.	Cytotherapy 18:1560-1563	2016	3,625	San Raffaele-Milano
PID	**Bernardo ME, Aiuti A**	The role of conditioning in hematopoietic stem cell gene therapy	Hum Gene Ther 27:741-748	2016	4,062	San Raffaele-Milano
CGD	Farinelli G, Jofra Hernandez R, Rossi A, Ranucci S, Sanvito F, Migliavacca M, Brombin C, Pramov A, Di Serio C, Bovolenta C, Gentner B, Bragonzi A, **Aiuti A**	Lentiviral Vector Gene Therapy Protects XCGD Mice From Acute Staphylococcus aureus Pneumonia and Inflammatory Response	Mol Ther, 24:1873-1880	2016	6,938	San Raffaele-Milano

PID	**Aiuti A, Naldini L**	Safer conditioning for blood stem cell transplants	Nat Biotechnol 34: 721-723	2016	43,113	San Raffaele-Milano
WAS	Biasco L, Pellin D, Scala S, Dionisio F, Basso-Ricci L, Leonardelli L, Scaramuzza S, Baricordi C, Ferrua F, Cicalese MP, Giannelli S, Neduva V, Dow DJ, Schmidt M, Von Kalle C, Roncarolo MG, Ciceri F, Vicard P, Wit E, Di Serio C, Naldini L, **Aiuti A**	In Vivo Tracking of Human Hematopoiesis Reveals Patterns of Clonal Dynamics during Early and Steady-State Reconstitution Phases.	Cell Stem Cell 19:107-119	2016	22,387	San Raffaele-Milano
ADA-SCID	**Cicalese MP**, Ferrua F, Castagnaro L, Pajno R, Barzaghi F, Giannelli S, Dionisio F, Brigida I, Bonopane M, Casiraghi M, Tabucchi A, Carlucci F, Grunebaum E, Adeli M, Bredius RG, Puck JM, Stepensky P, Tezcan I, Rolfe K, De Boever E, Reinhardt RR, Appleby J, Ciceri F, Roncarolo MG, **Aiuti A**	Update on the safety and efficacy of retroviral gene therapy for immunodeficiency due to adenosine deaminase deficiency.	Blood 128: 45-54	2016	11,847	San Raffaele-Milano
WAS	**Brigida I,** Scaramuzza S, Lazarevic D, Cittaro D, Ferrua F, Leonardelli L, Alessio M, Forma O, Lanzani C, Viarengo G, Ciceri F, Jankovic M, Pesce F, Aiuti A, **Cicalese MP.**	A novel genomic inversion in Wiskott-Aldrich-associated autoinflammation.	J Allergy Clin Immunol 138: 619-622	2016	12,485	San Raffaele-Milano
CGD	**Migliavacca M**, Assanelli A, Ferrua F, Cicalese MP, Biffi A, Frittoli M, Silvani P, Chidini G, Calderini E, Mandelli A, Camporesi A, Milani R, Farinelli G, Nicoletti R, Ciceri F, **Aiuti A, Bernardo ME**	Pioglitazone as a novel therapeutic approach in chronic granulomatous disease	J Allergy Clin Immunol, 137: 1913-1915.e.2	2016	12,485	San Raffaele-Milano

PID	Scarselli A, Di Cesare S, Capponi C, Cascioli S, Romiti ML, Di Matteo G, Simonetti A, Palma P, Finocchi A, Lucarelli B, Pinto RM, Rana I, Palumbo G, Caniglia M, Rossi P, Carsetti R, Cancrini C, **Aiuti A**	Longitudinal Evaluation of Immune Reconstitution and B-cell Function After Hematopoietic Cell Transplantation for Primary Immunodeficiency.	J Clin Immunol 35: 373-383	2015	3,094	San Raffaele-Milano
PID	**Cicalese MP, Aiuti A**	Clinical applications of gene therapy for primary immunodeficiencies	Human Gene Ther 26: 210-219	2015	6,938	San Raffaele-Milano
PID	Biasco L, Scala S, Basso Ricci L, Dionisio F, Baricordi C, Calabria A, Giannelli S, Cieri N, Barzaghi F, Pajno R, Al-Mousa H, Scarselli A, Cancrini C, Bordignon C, Roncarolo MG, Montini E, Bonini C, **Aiuti A**	In vivo tracking of T cells in humans unveils decade-long survival and activity of genetically modified T memory stem cells.	Sci Transl Med 7:273ra13	2015	16,264	San Raffaele-Milano
WAS	Bosticardo M, Ferrua F, Cavazzana M, **Aiuti A**	Gene therapy for Wiskott-Aldrich Syndrome	Curr Gene Ther 14:413-421	2014	2,542	San Raffaele-Milano
PID	Sauer AV, Di Lorenzo B, Carriglio N, **Aiuti A**	Progress in gene therapy for primary immunodeficiencies using lentiviral vectors.	Curr Opin Allergy Clin Immunol. 14:527-534	2014	N.A.	San Raffaele-Milano
X-CGD	Chiriaco M, Farinelli G, Capo V, Zonari E, Scaramuzza S, Di Matteo G, Sergi LS, Migliavacca M, Hernandez RJ, Bombelli F, Giorda E, Kajaste-Rudnitski A, Trono D, Grez M, Rossi P, Finocchi A, Naldini L, Gentner B, **Aiuti A**	Dual-regulated lentiviral vector for gene therapy of X-linked chronic granulomatosis	Mol Ther 22: 1472-1483	2014	6,227	San Raffaele-Milano

PID	Farinelli G, Capo V, Scaramuzza S, **Aiuti A.**	Lentiviral vectors for the treatment of primary immunodeficiencies	J Inherit Metab Dis. 37: 525-533	2014	3,365	San Raffaele-Milano
ADA	Brigida I, Sauer AV, Ferrua F, Giannelli S, Scaramuzza S, Pistoia V, Castiello MC, Barendregt BH, Cicalese MP, Casiraghi M, Brombin C, Puck J, Müller K, Notarangelo LD, Montin D, van Montfrans JM, Roncarolo MG, Traggiai E, van Dongen JJ, van der Burg M, **Aiuti A.**	B-cell development and functions and therapeutic options in adenosine deaminase-deficient patients.	J Allergy Clin Immunol. 133:799-806.e10	2014	11,476	San Raffaele-Milano
PID	**Martire B,** Panza R, Pillon M, Delvecchio M	CHARGE syndrome and common variable immunodeficiency: A case report and review of literature.	Pediatr Allergy Immunol. 27(5):546-50. IF 3,94	2016	3,94	Policlinico Bari
Ataxia-Teleangiectasia	**Chessa L**, Leuzzi V, Plebani A, Soresina A, Micheli R, D'Agnano D, Venturi T, Molinaro A, Fazzi E, Marini M, Ferremi Leali P, Quinti I, Cavaliere FM, Girelli G, Pietrogrande MC, Finocchi A, Tabolli S, Abeni D, Magnani M. .	Intra-erythrocyte infusion of dexamethasone reduces neurological symptoms in ataxia teleangiectasia patients: results of a phase 2 trial	Orphanet J Rare Dis. 9;9(1):5	2014	3,29	AO S.Andrea Roma
Agammaglobulinemia	**Lougaris V**, Baronio M, Vitali M, Tampella G, Cattalini M, Tassone L, Soresina A, Badolato R, **Plebani A**	Bruton tyrosine kinase mediates TLR9-dependent human dendritic cell activation	J Allergy Clin Immunol. pii: S0091-6749(14)00084-0	2014	11,2	Spedali Civili Brescia
CID	Caracciolo S, Moratto D, Giacomelli M, Negri S, Lougaris V, Porta F, Pajno G, Salpietro A, Montin D, Dinwiddie DL, Kingsmore SF, Plebani A, **Badolato R**	Expansion of CCR4+ activated T cells is associated with memory B cell reduction in DOCK8-deficient patients	Clin Immunol. 2 pii: S1521-6616(14)00072-2	2014	4.034	Spedali Civili Brescia

Agammaglobulinemia	**Lougaris V**, Vitali M, Baronio M, Moratto D, Tampella G, Biasini A, Badolato R, **Plebani A**	Autosomal recessive agammaglobulinemia: the third case of Igβ deficiency due to a novel non-sense mutation	J Clin Immunol. 34(4):425-7.	2014	3.094	Spedali Civili Brescia
PID	**Lougaris V**, Vitali M, Baronio M, Tampella G, **Plebani A**	BAFF-R mutations in Good's syndrome	Clin Immunol. 153(1):91-3.	2014	4.034	Spedali Civili Brescia
PID	Folsi V, Miglietti N, Lombardi A, Boccacci S, Utyatnikova T, Donati C, Squassabia L, Gazzola L, Bosio I, Borghi A, Grassi V, Notarangelo LD, **Plebani A**	Cardiomyopathy in a male patient with neutropenia and growth delay	Ital J Pediatr. 12;40:45.	2014	1.614	Spedali Civili Brescia
CID	**Lougaris V**, Baronio M, Vitali M, Gualdi G, Tampella G, Moratto D, Cattalini M, Pilotta A, Buzi F, Calzavara-Pinton P, **Plebani A**	Profound T-cell defects in Dubowitz syndrome.	Pediatr Allergy Immunol.25(5):511-3.	2014	3.947	Spedali Civili Brescia
SCG	Notarangelo L, Savoldi G, Cavagnini S, Bennato V, Vasile S, Pilotta A, **Plebani A, Porta F**.	Severe congenital neutropenia due to G6PC3 deficiency: early and delayed phenotype in two patients with two novel mutations	Ital J Pediatr. 14;40(1):80	2014	1.614	Spedali Civili Brescia
PID	**Badolato R**	Primary immunodeficiences: options for the future	Pediatr Allergy Immunol. 25(1):27-9	2014	3.947	Spedali Civili Brescia
CID	Dotta L, **Badolato R**	Primary immunodeficiencies appearing as combined lymphopenia, neutropenia, and monocytopenia	Immunol Lett. 161(2):222-5.	2014	2.483	Spedali Civili Brescia

CVID/IgA deficiency	Gualdi G, Lougaris V, Baronio M, Vitali M, Tampella G, Moratto D, Tanghetti P, Monari P, Calzavara-Pinton P, **Plebani A**	Burden of Skin Disease in Selective IgA Deficiency and Common Variable Immunodeficiency	J Investig Allergol Clin Immunol. 25(5):369-71	2015	2.131	Spedali Civili Brescia
APDS	**Lougaris** V, Faletra F, Lanzi G, Vozzi D, Marcuzzi A, Valencic E, Piscianz E, Bianco A, Girardelli M, Baronio M, Loganes C, Fasth A, Salvini F, Trizzino A, Moratto D, Facchetti F, Giliani S, Plebani A, Tommasini A	Altered germinal center reaction and abnormal B cell peripheral maturation in PI3KR1-mutated patients presenting with HIGM-like phenotype	Clin Immunol. 159: 33-6.	2015	4.034	Spedali Civili Brescia
CVID	**Lougaris V**, Ravelli A, Villanacci V, Salemme M, Soresina A, Fuoti M, Lanzarotto F, Lanzini A, Plebani A, Bassotti G	Gastrointestinal pathologic abnormalities in pediatric- and adult-onset common variable immunodeficiency	Dig Dis Sci. 60:2384-9.	2015	2.516	Spedali Civili Brescia
CVID	**Lougaris V**, Tabellini G, Vitali M, Baronio M, Patrizi O, Tampella G, Biasini A, Moratto D, Parolini S, **Plebani A**	Defective natural Killer-cell cytotoxic activity in NFKB2-mutated CVID-like disease.	J Allergy Clin Immunol. 135:1641-3	2015	11.200	Spedali Civili Brescia
Antibody deficiency	**Lougaris V**, Baronio M, Moratto D, Cardinale F, **Plebani A**	Monoallelic BAFFR P21R/H159Y mutations and familiar primary antibody deficiencies.	J Clin Immunol. 36(1):1-3	2016	3.094	Spedali Civili Brescia
CVID	**Lougaris V**, Baronio M, Masneri S, Lorenzini T, Cattivelli K, Tampella G, Soresina A, Moratto D, **Plebani A**	Correlation of bone marrow abnormalities, peripheral lymphocyte subsets and clinical features in uncomplicated common variable immunodeficiency (CVID) patients	Clin Immunol. 163:10-13	2016	4.034	Spedali Civili Brescia

PID	Dotta L, Scomodon O, Padoan R, Timpano S, Plebani A, Soresina A, Lougaris V, Concolino D, Nicoletti A, Giardino G, Licari A, Marseglia G, Pignata C, Tamassia N, Facchetti F, Vairo D, **Badolato R**	Clinical heterogeneity of dominant chronic mucocutaneous candidiasis disease: presenting as treatment-resistant candidiasis and chronic lung disease	Clin Immunol. 164:1-9	2016	4.034	Spedali Civili Brescia
PID	**Lougaris V**, Salpietro V, Cutrupi M, Baronio M, Moratto D, Pizzino MR, Mankad K, Briuglia S, Salpietro C, **Plebani A**	Proteus syndrome: evaluation of the immunological profile.	Orphanet J Rare Dis. 11:3	2016	3.290	Spedali Civili Brescia
22q11.2 deletion syndrome	Giacomelli M, Kumar R, Soresina A, Tamassia N, Lorenzini T, Moratto D, Gasperini S, Cassatella M, Plebani A, Lougaris V, **Badolato R**	Reduction of CRKL expression in patients with partial DiGeorge syndrome is associated with impairment of T-cell functions	J Allergy Clin Immunol. 138:229-240	2016	11.200	Spedali Civili Brescia
PID	Dotta L, Scomodon O, Padoan R, Timpano S, Plebani A, Soresina A, Lougaris V, Concolino D, Nicoletti A, Giardino G, Licari A, Marseglia G, Pignata C, Tamassia N, Facchetti F, Vairo D, **Badolato R**	Clinical and immunological data of nine patients with chronic mucocutaneous candidiasis disease.	Data Brief. 7:311-5.	2016		Spedali Civili Brescia
APDS	**Lougaris V**, Patrizi O, Baronio M, Tabellini G, Tampella G, Lanzi G, Salvini F, Trizzino A, Parolini S, **Plebani A**	p85α is an intrinsic regulator of human natural killer cell effector functions.	J Allergy Clin Immunol. 138:605-608	2016	11.200	Spedali Civili Brescia

PID	Prandini A, Salvi V, Colombo F, Moratto D, Lorenzi L, Vermi W, De Francesco MA, Notarangelo LD, Porta F, Plebani A, Facchetti F, Sozzani S, **Badolato R**	Impairment of dendritic cell functions in patients with adaptor protein-3 complex deficiency	Blood 127:3382-6.	2016	10.558	Spedali Civili Brescia
CVID	**Lougaris V**, Moratto D, Baronio M, Tampella G, van der Meer JW, Badolato R, Fliegauf M, **Plebani A**.	Early and late B-cell developmental impairment in nuclear factor kappa B, subunit 1-mutated common variable immunodeficiency disease	J Allergy Clin Immunol.	2016	11.200	Spedali Civili Brescia
CGD	Bondioni MP, Lougaris V, Di Gaetano G, Lorenzini T, Soresina A, Laffranchi F, Gatta D, **Plebani A**	Early Identification of Lung Fungal Infections in Chronic Granulomatous Disease (CGD) Using Multidetector Computer Tomography	J Clin Immunol	2016	3.094	Spedali Civili Brescia
PID	Lougaris V, Facchini E, Baronio M, Lorenzini T, Moratto D, Specchia F**, Plebani A**	Progressive severe B cell deficiency in pediatric Rubinstein-Taybi syndrome	Clin Immunol. 173:181-183.	2016	4.034	Spedali Civili Brescia
PID	**Lougaris V**, Patrizi O, Baronio M, Tabellini G, Tampella G, Damiati E, Frede N, van der Meer JW, Fliegauf M, Grimbacher B, Parolini S, **Plebani A**	NFKB1 regulates human NK cell maturation and effector functions	Clin Immunol. pii: S1521-6616(16)30170-X. doi: 10.1016/j.clim.2016.11.012	2016	4.034	Spedali Civili Brescia
Ataxia-Teleangiectasia	Piane M, Molinaro A, Soresina A, Costa S, Maffeis M, Germani A, Pinelli L, Meschini R, Plebani A, Chessa L, **Micheli R**	Novel compound heterozygous mutations in a child with Ataxia-Telangiectasia showing unrelated cerebellar disorders	J Neurol Sci. 15;371:48-53	2016		Spedali Civili Brescia

ALPS	Tripodi SI, Mazza C, Moratto D, Ramenghi U, Caorsi R, Gattorno M, **Badolato R**	Atypical presentation of autoimmune lymphoproliferative syndrome due to CASP10 mutation	Immunol Lett.177:22-4	2016	2.483	Spedali Civili Brescia
PID	Lorenzini T, Dotta L, Giacomelli M, Vairo D, **Badolato R**	STAT mutations as program switchers: turning primary immunodeficiencies into autoimmune diseases	J Leukoc Biol. pii: jlb.5RI0516-237RR.	2016	4.195	Spedali Civili Brescia
Agammaglobulinemia	Foiadelli T, Savasta S, Battistone A, Kota M, Passera C, Fiore S, Bino S, Amato C, Lozza A, Marseglia GL, **Fiore L**	Nucleotide variation in Sabin type 3 poliovirus from an Albanian infant with agammaglobulinemia and vaccine associatedpoliomyelitis	BMC Infect Dis.10;16:277	2016	2.690	IRCSS S. Matteo Pavia
PID	Tagliacarne SC, Valsecchi C, Castellazzi AM, Licari A, Klersy C, Montagna L,Castagnoli R, Benazzo M, Ciprandi G, **Marseglia GL.**	Impact of passive smokeand/or atopy on adenoid immunoglobulin production in children	Immunol Lett.165(2):70-7	2015	2.483	IRCSS S. Matteo Pavia
PID	Moscato GM, Di Matteo G, Ciotti M, Di Bonito P, Andreoni M, **Moschese V**	Dual response to human papilloma virus vaccine in an immunodeficiency disorder: resolution of plantar warts and persistence of condylomas	J Eur Acad Dermatol Venereol.30(7):1212-3	2016	3.029	Policlinico Tor Vergata
Antibody deficiency	**Moschese V**, Cavaliere FM, Graziani S, Bilotta C, Milito C, Chini L, **Quinti I**	Decreased IgM, IgA, and IgG response to pneumococcal vaccine in children with transient hypogammaglobulinemia of infancy.	J Allergy Clin Immunol. 137(2):617-9.	2016	12.485	Policlinico Tor Vergata
Autoimmunity	**Chini L,** Orlacchio A, Silenzi R, Della	Neuroimaging is useful for	Minerva Pediatr. 66(1):89-93	2014	0,532	Policlinico Tor

	Gatta F, Iannini R, Monteferrario E, Spano S, Guarino M, Moschese V, Simonetti G	monitoring disease activity in linear scleroderma "en coup de sabre".				Vergata
22q11.2 deletion syndrome	Cirillo E., Giardino G., Gallo V., Puliafito P., Azzari C., Bacchetta R., Cardinale F., Cicalese M.P., Consolini R., Martino S., Martire B., Molinatto C., Plebani A., Scarano G., Soresina A., Cancrini C., Rossi P., Digilio M.C., **Pignata C**	Intergenerational and intrafamilial phenotypic variability in 22q11.2 deletion syndrome subjects	BMC Med Genet. 15: 1.	2014	2.083	Policlinico Federico II
22q11.2 deletion syndrome	Giardino G., Cirillo E., Maio F., Gallo V., Esposito T., Naddei R., Grasso F., **Pignata C.**	Gastrointestinal involvement in patients affected with 22q11.2 deletion syndrome	Scand J Gastroenterol.49: 247-79.	2014	2.361	Policlinico Federico II
SCID	Palamaro L., Romano R., Fusco A., Giardino G., Gallo V., **Pignata C**	FOXN1 in cell development and human diseases	Int Rev Immunol. 33: 83-93.	2014	4.103	Policlinico Federico II
SCID	**Pignata C.**, Romano R	FOX genes and the immune response.	Int Rev Immunol. 33: 81-2	2014	4.103	Policlinico Federico II
PID	D'Assante R., Fusco A., Palamaro L., Giardino G., Gallo V., Cirillo E., **Pignata C.**	Unraveling the link between ectodermal disorders and primary immunodeficiencies	Int Rev Immunol. 35: 25-38	2015	4.103	Policlinico Federico II
PID	**Pignata C.**, D'Assante R., Sousa AE	Thymic stromal alterations and genetic disorders of immune system.	Front Immunol. 6: 81, 2015	2015		Policlinico Federico II
SCID	Cirillo E., Giardino G., Gallo V., D'Assante R., Grasso F., Romano R., Di Lillo C., Galasso G., **Pignata C**	Severe combined immunodeficiency-an update.	Ann N Y Acad Sci. 1356: 90-106	2015	4.383	Policlinico Federico II
PID	Giardino G., Cirillo E., Gallo V., Esposito T., Fusco F., Conte M.I.,	B cells from Nuclear factor kB essential modulator deficient	Clin Immunol. 161: 131-5	2015	3.672	Policlinico

	Quinti I., Ursini MV., Carsetti R., **Pignata C**	patients fail to differentiate to antibody secreting cells in response to TLR9 ligand				Federico II
PID	Giardino G., Gallo V., Somma D., Farrow E.G., Thiffault I., D'Assante R., Donofrio V., Paciolla M., Ursini MV., Leonardi A., Saunders C.J., **Pignata C**	Targeted next generation sequencing revealed MYD88 deficiency in a child with chronic Yersiniosis and granulomatous lymphadenitis	J All Clin Immunol. 137: 1591-5	2015	11.476	Policlinico Federico II
PID	Giardino G., Somma D., Cirillo E., Ruggiero G., Terrazzano G., Rubino V., Ursini M.V., Vairo D., Badolato R., Carsetti R., Leonardi A., Puel A., **Pignata C**	Novel STAT1 gain-of-function mutation and suppurative infections	Pediatr Allergy Immunol. 27: 220-3,	2016	3.397	Policlinico Federico II
22q11.2 deletion syndrome	Romano R., Cirillo, E., Giardino G., Gallo V., Mollica C., **Pignata C**	A broncho-vascular anomaly in a patient with 22q11.2 deletion syndrome.	J Investig Allergol Clin Immunol. 26: 390-2	2016	2.596	Policlinico Federico II
Immunodeficiency and autoimmunity	Giardino G., Gallo V., Prencipe R., Gaudino G., Romano R., De Cataldis M., Lorello, P., Palamaro L., Di Giacomo C., Capalbo D., Cirillo E., D'Assante R., **Pignata C**	Unbalanced immune system: immunodeficiencies and autoimmunity	Front Pediatr. 4: 107	2016		Policlinico Federico II
PID	Gallo V., Dotta L., GiardinoG., Cirillo E., Lougaris V., D'AssanteR., PrandiniA., ConsoliniR., Farrow E.G., ThiffaultI., Saunders C.J., LeonardiA., PlebaniA., BadolatoR., **Pignata C**	Diagnostics of Primary Immunodeficiencies through Next Generation Sequencing.	Front Immunol. 7: 466.	2016	5.695	Policlinico Federico II

Ataxia-teleangiectasia	D'Assante R., Fusco A., Palamaro L., Polishchuk E., Polishchuk R., Grieco V., Prencipe M.R., Ballabio A., Pignata C	Abnormal cell-clearance and accumulation of autophagic vesicles in lymphocytes from patients affected with Ataxia-Teleangiectasia	Clin Immunol. 175: 16-25	2016	4.034	Policlinico Federico II
PID	Canessa C, Iacopelli J, Pecoraro A, Spadaro G, Matucci A, Milito C, Vultaggio A, Agostini C, Cinetto F, Danieli MG, Gambini S, Marasco C, Trizzino A, Vacca A, De Mattia D, Martire B, Plebani A, Di Gioacchino M, Gatta A, Finocchi A, Licciardi F, Martino S, De Carli M, Moschese V, **Azzari C**	Shift from intravenous or 16% subcutaneous replacement therapy to 20% subcutaneous immunoglobulin in patients with primary antibody deficiencies	Int J Immunopathol Pharmacol	2017	2.347	Ospedale Meyer Firenze
PID	Ricci S, Romano F, Nieddu F, Picard C, **Azzari C**	OL-EDA-ID Syndrome: a Novel Hypomorphic NEMO Mutation Associated with a Severe Clinical Presentation and Transient HLH.	J Clin Immunol	2017	3.253	Ospedale Meyer Firenze
PID	la Marca G, Canessa C, Giocaliere E, Romano F, Malvagia S, Funghini S, Moriondo M, Valleriani C, Lippi F, Ombrone D, Della Bona ML, Speckmann C, Borte S, Brodszki N, Gennery AR, Weinacht K, Celmeli F, Pagel J, de Martino M, Guerrini R, Wittkowski H, Santisteban I, Bali P, Ikinciogullari A, Hershfield M, Notarangelo LD, Resti M, **Azzari C**	Diagnosis of immunodeficiency caused by a purine nucleoside phosphorylase defect by using tandem mass spectrometry on dried blood spots	J Allergy Clin Immunol	2014	11.476	Ospedale Meyer Firenze

SCID	la Marca G, Giocaliere E, Malvagia S, Funghini S, Ombrone D, Della Bona ML, Canessa C, Lippi F, Romano F, Guerrini R, Resti M, **Azzari C.**	The inclusion of ADA-SCID in expanded newborn screening by tandem mass spectrometry	J Pharm Biomed Anal	2014	2.979	Ospedale Meyer Firenze

Trial clinici

Novel drugs under development for rare immune disorders

Disease	Drug	Company	Centers in Italy	ODD and clinical trial
Ataxia telangectasia (ATM)	Dexamethasone sodium phosphate encapsulated in human autologous erythrocytes (Erydex)	Erydel	Spedali Civili Brescia- Università di Roma La Sapienza	USA and EU/ Phase III

Novel drugs under development for rare immune disorders

Disease	Drug	Company	Centers in Italy	ODD and clinical trial
Hemophagocytic Lymphohistiocytosis	Recombinant human anti-interferon gamma monoclonal antibody (Emapalumab NI-0501)	Novimmune SA	OPBG (Rome) Gaslini (Genova) Meyer (Firenze) San Gerardo (Monza) Verona	USA and EU/ Phase II

Novel drugs under development for rare immune disorders

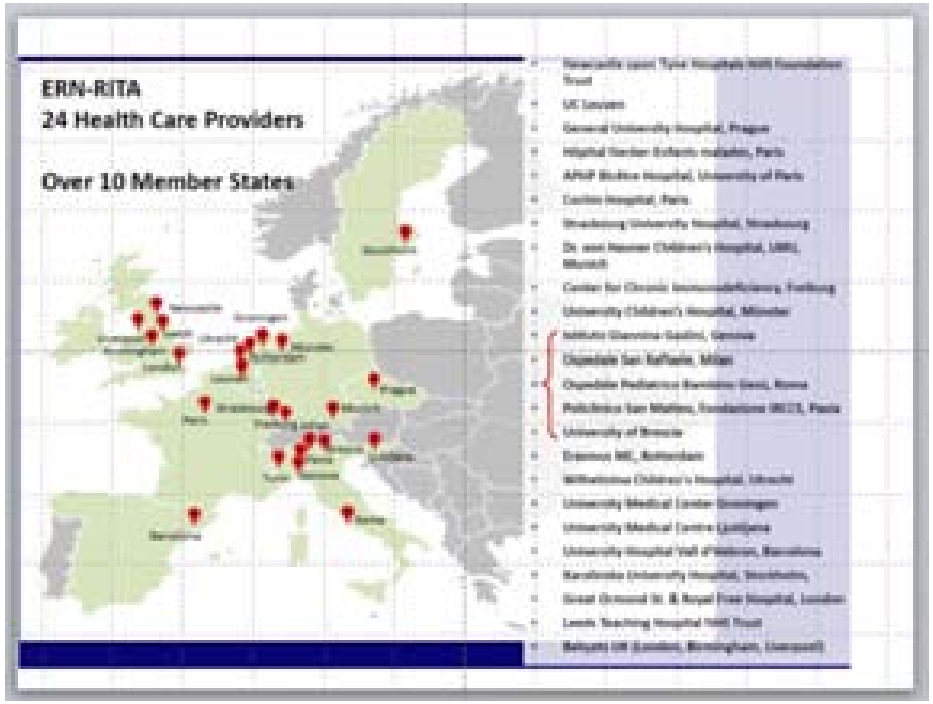
Disease	Drug	Company	Center	Notes
Activated phosphoinositide 3 kinase (pi3k) delta syndrome (aods)	UCB5857	UCB	OPBG (Cancrini)	Phase II
Activated phosphoinositide 3 kinase (pi3k) delta syndrome (aods)	CDZ173	Novartis	Spedali Civili Brescia	
WHIM Syndrome (CXCR4 deficiency)	X4P-001	X4 Pharmaceuticals	Spedali Civili Brescia	Phase II Phase III

Novel drugs under development for rare immune disorders

Disease	Drug	Company	Center	Odd /Study
Various conditions (including PID)	Allogeneic donor-derived T cells lymphocytes transduced with inducible caspase 9 /CD19 (BPX-501)	Bellicum	OPBG (Rome)	ODD in EU and US Phase II
ADA-SCID	STRIMVELIS (autologous gene therapy)	GSK	SR-TIGET (Milan)	ODD EU and US Approved drug in the EU
Wiskott-Aldrich Syndrome	Autologous LV gene therapy	GSK	SR-TIGET (Milan)	ODD EU and US Phase I/II

Partecipazione Nazionale a ERN

La ricognizione dei centri pediatrici che sono stati accettati nella rete europea di riferimento (ERN) per le malattie immunologiche pediatriche



Fibrosi cistica

A cura della Società Italiana per lo studio della Fibrosi Cistica – SIFC
(Presidente: *Valeria Raia*)

Valeria Raia

Pubblicazioni, *Trial* clinici farmacologici, e Partecipazione Nazionale a ERN

Valeria Raia
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Responsabile Centro Regionale di Riferimento della Regione Campania Unità Pediatrica
Presidente Società Italiana per lo studio della Fibrosi Cistica

La Fibrosi Cistica è una malattia genetica, cronica, ad alta complessità di gestione, dove la storia naturale della malattia è correlata alla continuità e alla qualità delle cure. Per questo, pur presentando la Fibrosi Cistica caratteristiche di malattia rara, è tutelata dalla Legge Nazionale 548/93 che prevede, fra l'altro, l'istituzione di centri di Riferimento Specializzati in tutte le Regioni con funzioni di prevenzione, diagnosi e cura della malattia e, dove esistano le condizioni adeguate, di ricerca.

I dati epidemiologici inclusi nel Registro Italiano di Fibrosi Cistica (www.registroitalianofibrosicistica.it) aggiornati al 2014, confrontati con quelli riportati a livello europeo ed internazionale, dimostrano che i modelli di cura attuati in Italia sono adeguati, nonostante le numerose difficoltà di tipo organizzativo e sanitario, con più del 50% di malattia viventi oltre i 18 anni. I dati incoraggianti degli ultimi anni provenienti dalla ricerca e dalla identificazione di nuove terapie in grado, almeno per il momento, di stabilizzare l'evoluzione della malattia legittimano le speranze per un futuro sempre migliore ed una sopravvivenza sempre più lunga. Per questo, se da una parte si conferma la necessità di incrementare studi clinici controllati in grado di verificare l'efficacia e la sicurezza di nuovi farmaci che prepotentemente si affacciano nel panorama della Fibrosi Cistica, dall'altra il modello di medicina traslazionale può rappresentare oggi il vero collante tra la ricerca biologica preclinica e la ricerca clinica, trasferendo in modo rapido le nuove conoscenze dalla scienza di base a quella biomedica. Le diverse competenze che operano nel campo della Fibrosi Cistica (epidemiologia, gastroenterologia, pneumologia, infettivologia, genetica, microbiologia) impongono un continuo scambio fra ricercatori dedicati prevalentemente alla clinica e quelli orientati alla ricerca biologica che vede prestigiosi laboratori italiani orientati a diverse aree di sviluppo scientifico.

Tuttavia, motivi etici e caratteristiche di evolutività della malattia rendono difficile la organizzazione di studi clinici controllati, con possibili rischi di *bias* nella metodologia applicata e nella interpretazione dei risultati. Per questo, il potenziale di nuove terapie in grado di modulare il difetto genetico di base della malattia apre lo scenario al modello della medicina di precisione sulla base di interventi individualizzati.

L'affiliazione alle Reti di Riferimento Europee (ERN-LUNG) di alcuni Centri di Riferimento per la cura della Fibrosi Cistica incardinati nelle rispettive strutture Aziendali/Universitarie (Azienda Ospedaliera Universitaria Policlinico, Università degli Studi di Napoli Federico II, Napoli; Ospedale Pediatrico Bambino Gesù, Roma; Azienda Ospedaliero Universitaria Meyer, Firenze; Azienda Ospedaliera Universitaria Integrata, Verona) conferma la disponibilità e la competenza per una maggiore qualificazione e supporto alla ricerca clinica non solo in Italia, ma anche a livello europeo.

Pubblicazioni

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Fibrosi Cistica	Terlizzi V, Castaldo G, Salvatore D, Lucarelli M, Raia V, Angioni A, Carnovale V, Cirilli N, Casciaro R, Colombo C, Di Lullo AM, Elce A, Iacotucci P, Comegna M, Scorza M, Lucidi V, Perfetti A, Cimino R, Quattrucci S, Seia M, Sofia VM, Zarrilli F, Amato F.	Genotype-phenotype correlation and functional studies in patients with cystic fibrosis bearing CFTR complex alleles.	Journal of Medical Genetics 54(4):224-235. doi: 10.1136/jmedgenet-2016-103985.	2017	5.650	Collaboration
Fibrosi Cistica	Lucarelli M	New era of cystic fibrosis: full mutational analysis and personalized therapy.	World Journal of Medical Genetics 7(1):1-9. doi: 10.5496/wjmg.v7.i1.1	2017	0	Invited editorial (no Pubmed)
Fibrosi Cistica	Lucarelli M, Bruno SM, Pierandrei S, Ferraguti G, Testino G, Truglio G, Strom R, Quattrucci S	The impact on genetic testing of mutational patterns of CFTR gene in different clinical macrocategories of cystic fibrosis.	Journal of Molecular Diagnostics 18(4):554-565. doi: 10.1016/j.jmoldx.2016.02.007.	2016	5.201	
Fibrosi Cistica	Lucarelli M	Commentary on “The novel complex allele [A238V;F508del] of the CFTR gene: clinical phenotype and possible implications for cystic fibrosis etiological therapies”.	Journal of Human Genetics 61(6):471-472. doi: 10.1038/jhg.2016.34.	2016	2.487	Invited commentary

Fibrosi Cistica	Lucarelli M, Bruno SM, Pierandrei S, Ferraguti G, Stamato A, Narzi F, Amato A, Cimino G, Bertasi S,Quattrucci S, Strom R	A genotypic-oriented view of CFTR genetics highlights specific mutational patterns underlying clinical Macro-categories of cystic fibrosis.	Molecular Medicine21:257-275. doi: 10.2119/molmed.2014.00229	2015	3.530	
Fibrosi Cistica	Terlizzi V, Carnovale V, Castaldo G, Castellani C, Cirilli N, Colombo C, Corti F, Cresta F, D'Adda A, Lucarelli M, Lucidi V, Macchiaroli A, Madarena E, Padoan R, Quattrucci S, Salvatore D, Zarrilli F, Raia V	Clinical expression of patients with the D1152H CFTR mutation.	Journal of Cystic Fibrosis 14:447-452. doi: 10.1016/j.jcf.2014.12.012.	2015	3.853	Collaboration
Fibrosi Cistica	Savi D, Simmonds N, Di Paolo M, Quattrucci S, Palange P, Banya W, Hopkinson NS, Bilton D	Relationship between pulmonary exacerbations and daily physical activity in adults with cystic fibrosis.	BMC Pulmonary Medicine 15:151. doi: 10.1186/s12890-015-0151-7	2015	2.329	
Fibrosi Cistica	Nebbioso M, Quattrucci S, Leggieri E, Spadea L, Vingolo EM.	Cystic fibrosis and new trends by ophthalmological evaluation: a pilot study.	BioMed Research International :580373. doi: 10.1155/2014/580373.	2014	1.579	
Fibrosi Cistica	Iebba V, Totino V, Santangelo F, Gagliardi A, Ciotoli L, Virga A, Ambrosi C, Pompili M, De Biase RV, Selan L, Artini M, Pantanella F, Mura F, Passariello C, Nicoletti M, Nencioni L, TrancassiniM,Quattrucci S, Schippa S.	Bdellovibrio bacteriovorus directly attacks Pseudomonas aeruginosa and Staphylococcus aureus cystic fibrosis isolates.	Frontiers in Microbiology 5:280. doi: 10.3389/fmicb.2014.00280.	2014	4.165	

Fibrosi Cistica	Savi D, De Biase RV, Amaddeo A, Anile M, Venuta F, Ruberto F, Simmonds N, Cimino G, Quattrucci S.	Burkholderia pyrrocinia in cystic fibrosis lung transplantation: a case report.	Transplantation Proceedings 46(1):295-7. doi:10.1016/j.transproceed.2013.08.108	2014	0.982	
Fibrosi Cistica	Crisafulli E, Frizzelli A , Fantin A, Manco A, Mangia A, Pisi G, Fainardi V, Aiello M, Bertorelli G, Chetta A.	Next generation beta adrenoreceptor agonists for the treatment of asthma	Expert Opinion On Pharmacotherapy	2017	3.543	In press
Fibrosi Cistica	Crisafulli E, Zanini A, Pisi G, Pignatti P, Poli G, Scuri M, Chetta A.	Inhaled beclometasone dipropionate/formoterol fumarate extrafine fixed combination for the treatment of asthma.	Expert Rev Respir Med. 10(5):481-90.	2016	2.1	
Fibrosi Cistica	Montanini L, Smerieri A, Gullì M, Cirillo F, Pisi G, Sartori C, Amarri S, Bernasconi S, Marmiroli N, Street ME.	MiR-146a, miR-155, miR-370 and miR-708 are CFTR-dependent, Predicted FOXO1 Regulators and Change at Onset of CFRDs.	J Clin Endocrinol Metab. 101(12):4955-4963	2016	5.5	
Fibrosi Cistica	Montanini L, Cirillo F, Smerieri A, Pisi G, Giardino I, d'Apolito M, Spaggiari C, Bernasconi S, Amarri S, Street ME.	HMGB1 Is Increased by CFTR Loss of Function, Is Lowered by Insulin, and Increases In Vivo at Onset of CFRD.	J Clin Endocrinol Metab. 101(3):1274-81	2016	5.5	
Fibrosi Cistica	Fainardi V, Pisi G, Chetta A.	Mepolizumab in the treatment of severe eosinophilic asthma.	Immunotherapy 8(1):27-34.	2016	2.0	
Fibrosi Cistica	Taccetti G, Sly PD.	Early detection of infection with Pseudomonas aeruginosa in cystic fibrosis: the Holy Grail or an achievable goal?	J Cyst Fibros. Sep;13(5):491-3	2014	3.475	

Fibrosi Cistica	Dolce D, Cariani L, Campana S, Ravenni N, Mergni G, Biffi A, Colombo C, Gagliardini R, Cirilli N, Manso E, Padoan R, Soncini E, Forte FR, D'Aprile A, Ratclif L, Amboni M, Casciaro R, Minicucci L, Borio T, Cosimi A, Vieni G, Zinnarello C, Fiscarelli E, Collura M, Pensabene T, Braggion C, Döring G, Taccetti G.	Evaluation of specific immune response in early P. aeruginosa infection in cystic fibrosis patients.	J Cyst Fibros. Jan;13(1):116-7	2014	3.475	Collaboration
Fibrosi Cistica	Buzzetti R, Cirilli N, Minicucci L, Raia V, Salvatore D, Maffeis P.	Cystic Fibrosis Database (CFDB): A new web-based tool for cystic fibrosis specialists	Pediatr Pulmonol. Sep;49(9):938-40	2014	2.704	
Fibrosi Cistica	Salvatore M, Floridia G, Amato A, Censi F, Carta C, de Stefano MC, Ferrari G, Tosto F, Capoluongo E, Caruso U, Castaldo G, Cirilli N, Corbetta C, Padoan R, Raia V, Taruscio D.	The Italian pilot external quality assessment program for cystic fibrosis sweat test	Clinical Biochemistry;49(7):601-5	2016	2.382	
Fibrosi Cistica	Maglione M, Montella S, Mollica C, Carnovale V, Iacotucci P, De Gregorio F, Tosco A, Cervasio M, Raia V, Santamaria F.	Lung structure and function similarities between primary ciliary dyskinesia and mild cystic fibrosis: a pilot study.	Ital J Pediatr. Apr 12;43(1):34.	2017	1.614	

Fibrosi Cistica	Ferrari E, Monzani R, Villella VR, Esposito S, Saluzzo F, Rossin F, D'Eletto M, Tosco A, De Gregorio F, Izzo V, Maiuri MC, Kroemer G, Raia V, Maiuri L.	Cysteamine re-establishes the clearance of Pseudomonas aeruginosa by macrophages bearing the cystic fibrosis-relevant F508del-CFTR mutation.	Cell Death Dis. Jan 12;8(1):e2544.	2017	5.378	
Fibrosi Cistica	Tosco A, De Gregorio F, Esposito S, De Stefano D, Sana I, Ferrari E, Sepe A, Salvadori L, Buonpensiero P, Di Pasqua A, Grassia R, Leone CA, Guido S, De Rosa G, Lusa S, Bona G, Stoll G, Maiuri MC, Mehta A, Kroemer G, Maiuri L, Raia V.	A novel treatment of cystic fibrosis acting on-target: cysteamine plus epigallocatechin gallate for the autophagy-dependent rescue of class II-mutated CFTR.	Cell Death Differ. Aug;23(8):1380-93.	2016	8.128	
Fibrosi Cistica	Maiuri L, De Stefano D, Raia V, Kroemer G.	The holy grail of cystic fibrosis research: pharmacological repair of the F508del-CFTR mutation.	Ann Transl Med. May;3(Suppl 1):S24.	2015		
Fibrosi Cistica	Franzese A, Mozzillo E, Fattorusso V, Raia V, Valerio G.	Screening of glucose metabolism derangements in pediatric cystic fibrosis patients: how, when, why.	Acta Diabetol. Aug;52(4):633-8.	2015	3.074	
Fibrosi Cistica	Calabrese C, Tosco A, Abete P, Carnovale V, Basile C, Magliocca A, Quattrucci S, De Sanctis S, Alatri F, Mazzarella G, De Pietro L, Turino C, Melillo E, Buonpensiero P, Di Pasqua A, Raia V.	Randomized, single blind, controlled trial of inhaled glutathione vs placebo in patients with cystic fibrosis.	J Cyst Fibros. Mar;14(2):203-10	2015	3.853	

Fibrosi Cistica	De Stefano D, Villella VR, Esposito S, Tosco A, Sepe A, De Gregorio F, Salvadori L, Grassia R, Leone CA, De Rosa G, Maiuri MC, Pettoello-Mantovani M, Guido S, Bossi A, Zolin A, Venerando A, Pinna LA, Mehta A, Bona G, Kroemer G, Maiuri L, Raia V.	Restoration of CFTR function in patients with cystic fibrosis carrying the F508del-CFTR mutation.	Autophagy. 10(11):2053-74	2014	11.753	
Fibrosi Cistica	Terlizzi V, Tosco A, Tomaiuolo R, Sepe A, Amato N, Casale A, Mercogliano C, De Gregorio F, Improta F, Elce A, Castaldo G, Raia V.	Prediction of acute pancreatitis risk based on PIP score in children with cystic fibrosis.	J Cyst Fibros. Sep;13(5):579-84	2014	3.475	
Fibrosi Cistica	Bruzzese E, Callegari ML, Raia V, Viscovo S, Scotto R, Ferrari S, Morelli L, Buccigrossi V, Lo Vecchio A, Ruberto E, Guarino A.	Disrupted intestinal microbiota and intestinal inflammation in children with cystic fibrosis and its restoration with Lactobacillus GG: a randomised clinical trial.	PLoS One. Feb 19;9(2):e87796.	2014	4.17	
Fibrosi Cistica	Veropalumbo C, Campanozzi A, De Gregorio F, Correra A, Raia V, Vajro P.	Shwachman-Diamond syndrome with autoimmune-like liver disease and enteropathy mimicking celiac disease.	Clin Res Hepatol Gastroenterol. Feb;39(1):e1-4	2015	1.872	
Fibrosi Cistica	Terlizzi V, Zito E, Mozzillo E, Raia V, Franzese A.	Can continuous subcutaneous insulin infusion improve health-related quality of life in patients with Shwachman-Bodian-Diamond syndrome and diabetes?	Diabetes Technol Ther. Jan;17(1):64-7	2015	2.198	

Trial clinici farmacologici

1 **Nome MR**	2 ***Sponsor***	3 ***Partners***		4 **Ruolo**		5 **Denominazione del *trial***	6 **Farmaco**	7 **Durata**		8 **Note**
		I	**N**	**C**	**P**			**C**	**T (anno)**	
Fibrosi Cistica	Anthera Pharmaceuticals	X			X	"A phase III pivotal study in patients with Cystic Fibrosis to evaluate a soluble, non-porcine, stable, pancreatic enzyme replacement product (liprotamase) for the treatment of exocrine pancreatic insufficiency"	liprotamase		X 2015	Multicentrico
Fibrosi Cistica	Forest	X			X	"A prospective parallel group comparison of Colobreathe, and tobramycin nebulizer solution in the management of early lung infection with pseudomonas aeruginosa in paediatric patients with CysticFibrosis".	Colobreathe, tobramycin		X 2014	Multicentrico
Fibrosi Cistica	Vertex Pharmaceuticals	X			X	VX BOI CF	-		X 2015	Multicentrico
Fibrosi Cistica	Vertex Pharmaceuticals	X			X	VX14-CFR-107	-	X		Multicentrico

Fibrosi Cistica	Vertex Pharmaceuticals	X			X	VX12-809- 103 “A Phase 3, Randomized, Double-Blind, Placebo-Controlled, Parallel-Group Study to Evaluate the Efficacy and Safety of Lumacaftor in Combination With Ivacaftor in Subjects Aged 12 Years and Older With Cystic Fibrosis, Homozygous for the F508del-CFTR Mutation”	VX770+VX809		X 2014	Multicentrico con Centro Coordinatore italiano a Milano
Fibrosi Cistica	Vertex Pharmaceuticals	X			X	VX12809105 "A Phase 3, Rollover Study to Evaluate the Safety and Efficacy of Long-term Treatment With Lumacaftor in Combination With Ivacaftor in Subjects Aged 12 Years and Older With Cystic Fibrosis, Homozygous or Heterozygous for the F508del-CFTR Mutation"	VX770+VX809		X 2016	Multicentrico
Fibrosi Cistica	Vertex Pharmaceuticals	X			X	VX14-661-106 "A Randomized, double blind, placebo controlled, parallel group study to evaluate the efficacy and safety of VX 661 in combination with ivacaftor".	VX770+VX 661		X 2016	Multicentrico

Fibrosi Cistica	Vertex Pharmaceuticals	X			X	VX14-661-109 "A phase 3 study of VX-661 in Combination with Ivacaftor in subjects aged 12 years and older with Cystic Fibrosis, who have one F508del-CFTR mutation and a second mutation that has been demonstrated to be clinically responsive to ivacaftor."	VX770+VX 661		X 2016	Multicentrico
Fibrosi Cistica	Vertex Pharmaceuticals	X			X	VX14-661-108 "A Phase 3, Randomized, Double-Blind, Placebo-Controlled, Crossover Study to Evaluate the Efficacy and Safety of Ivacaftor and VX-661 in Combination With Ivacaftor in Subjects Aged 12 Years and Older With Cystic Fibrosis, Heterozygous for the F508del-CFTR Mutation, and a Second Allele With a CFTR Mutation Predicted to Have Residual Function ."	VX770+VX 661		X 2016	Multicentrico

Fibrosi Cistica	Vertex Pharmaceuticals	X			X	A Phase 3, Open-label, Rollover Study to Evaluate the Safety and Efficacy of Long-term Treatment With VX-661 in Combination With Ivacaftor in Subjects Aged 12 Years and Older With Cystic Fibrosis, Homozygous or Heterozygous for the *F508del-CFTR* Mutation Vertex (VX14-661-110)	VX 661-VX770	X		Multicentrico
Fibrosi Cistica	Novartis Farma	X			X	CTBM100C2401 Studio a singolo braccio, in aperto, multicentrico, di fase IV per valutare la sicurezza a lungo termine della tobramicina in polvere per inalazione (TIP) in pazienti affetti da Fibrosi Cistica.	Tobi Podahaler		X 2014	Multicentrico
Fibrosi Cistica	Aptalis	X			X	Randomised, Double-Blind, Active-Controlled, Two-Treatment, Crossover, Multinational, Multicentre Study to Compare Two Pancreatic Enzyme Products in the Treatment of Exocrine Pancreatic Insufficiency in Subjects With Cystic Fibrosis PR005	Pancreatic Enzyme		X 2014	Multicentrico

Fibrosi Cistica			X	X		Studio clinico pilota di ricerca sperimentale di fase II, per valutare il recupero funzionale della proteina CFTR attraverso l'uso di regolatori della proteostasi.	Cysteamina + EGCG		X 2015	Monocentrico
Fibrosi Cistica	NovaBiotics	X			X	A study of the dosing, efficacy, and safety of Oral Cysteamine in adult patient with Cystic Fibrosis Exacerbations (CARE-CF1)	Cysteamine	X		Multicentrico
Fibrosi Cistica	AIFA		X	X		FARM 7K7XZB Inhaled glutathione (GSH) versus Placebo in Cystic Fibrosis	Glutathione		X 2014	Monocentrico
Fibrosi Cistica	Pharmaxis Limited	X			X	DPM-CF-303 "A safety and efficacy trial of inhaled mannitol in adult Cystic Fibrosis Subjects"	mannitol		X 2017	Multicentrico
Fibrosi Cistica	Pharmaxis Limited	X			X	DPM-CF 204 "Crossover trial determining the efficacy of dry powder mannitol to improve lung function in subjects aged 6-17 years"	mannitol		X 2014	Multicentrico
Fibrosi Cistica	Novartis Farma	X			X	CTBM100C2409	Tobi		X 2015	Multicentrico

Fibrosi Cistica	Chiesi farmaceutici		X		X	Studio osservazionale prospettico a lungo termine sulla sicurezza e tollerabilità di Bramitob somministrato due volte al giorno per tre cicli di 28 giorni on/28 giorni off, in pazienti con fibrosi cistica aventi funzionalità polmonare severamente compromessa".	Bramitob	X		Multicentrico Centro Coordinatore Parma
Fibrosi Cistica	Impactt	X			X	PsAer-IgY: "Efficacy study of IgY (Antibody Against Pseudomonas) in Cystic Fibrosis Patients (PsAer-IgY)	Avian polyclonal anti-Pseudomonas antibodies (IgY)		X 2016	Multicentrico
Fibrosi Cistica	PTC Therapeutics	X			X	PTC124-GD-023-CF "Study of Ataluren in Cystic Fibrosis"	PTC 124	X		Multicentrico
Fibrosi Cistica	PTC Therapeutics	X			X	A phase 3 efficacy and safety study of Ataluren (PTC124®) in patients with nonsense mutation cystic fibrosis (Protocol Number PTC124-GD-021-CF)	PTC 124		X 2016	Multicentrico

Fibrosi Cistica	PTC Therapeutics	X			X	A PHASE 3 EXTENSION STUDY OF ATALUREN (PTC124) IN PATIENTS WITH NONSENSE MUTATION CYSTIC FIBROSIS (PTC124-GD-021e-CF)A PHASE 3 EXTENSION STUDY OF ATALUREN (PTC124) IN	PTC 124		X 2017	Multicentrico
Fibrosi Cistica	Celtaxsys	X			X	A Phase 2, Multicenter, Randomized, Double-blind, Placebo-controlled, Parallel-group Study to Evaluate the Efficacy, Safety, and Tolerability of CTX-4430 Administered Orally Once-Daily for 48 Weeks in Adult Patients with Cystic Fibrosis (CTX-4430-CF-201)	CTX-4430	X		Multicentrico
Fibrosi Cistica	Fondazione Fibrosi Cistica		X		X	FFC #30/2015 "Studio randomizzato multicentrico sull'eradicazione di Pseudomonas aeruginosa in pazienti con fibrosi cistica: confronto tra il trattamento eradicante classico e il trattamento classico associato con la terapia antibiotica delle alte vie respiratorie"		X		Multicentrico Firenze Centro Coordinatore

Malattie respiratorie infantili

A cura della Società Italiana per le Malattie Respiratorie Infantili – SIMRI
(Presidente: *Renato Cutrera*)

Francesca Santamaria

Pubblicazioni, *Trial* clinici farmacologici, e Partecipazione Nazionale a ERN

Francesca Santamaria
Rappresentante dell'Health Care Provider AOU Federico II di Napoli per l'ERN Lung

Relativamente all'attività in tema di pubblicazioni e *trial* clinici nel settore delle malattie polmonari rare, i dati inseriti nel database sono stati ottenuti attraverso una ricerca sulla piattaforma PubMed, inserendo quali parole chiave i nominativi delle varie patologie rare polmonari (siano esse inserite e non nell'elenco delle malattie rare fornito dal Ministero della Salute italiano) and Italy and children and/or adolescents, e selezionando le pubblicazioni inerenti con primo/ultimo nome o corresponding author un autore afferente ad un centro italiano. La ricerca è stata chiusa al 28 giugno 2017.
Gli inserimenti nel database sono distribuiti nelle seguenti categorie:

- *Congenital lung malformations*
- *Interstitial Lung Disease*
- *Primary ciliary dyskinesia*
- *Airways cancer*
- *Congenital Chronic Hypoventilation Syndrome*
- *Non- Cystic Fibrosis bronchiectasis*
- *Lung transplant*

Considerando le singole patologie, le pubblicazioni più numerose sono state osservate nell'ambito della discinesia ciliare primitiva, seguite dalla produzione nell'ambito delle pneumopatie interstiziali e delle malformazioni polmonari congenite. Per quanto concerne la qualità delle pubblicazioni, il range dell'impact factor è 0,7-13.
Solo una minoranza delle pubblicazioni incluse nel database sono il risultato di lavori cooperativi sia italiani che internazionali.
Pochi sono i trial clinici nel settore delle malattie polmonari rare in età pediatrica, di questi uno solo riguarda un farmaco "orfano", mentre un trial in corso in due diversi centri italiani è relativo ad un farmaco non "orfano", ma il cui uso non è ancora codificato.

La "**European Reference Network on lung diseases"** (ERN Lung), coordinata dal Prof. Thomas Wagner, Università di Francoforte, comprende nove *Core Networks*:

1. Pneumopatie Interstiziali
2. Fibrosi Cistica
3. Discinesia Ciliare Primitiva
4. Ipertensione Polmonare
5. Bronchiettasie Non- Fibrosi Cistica
6. Deficit di alfa1-antitripsina
7. Mesotelioma
8. Disfunzione cronica da rigetto polmonare
9. Altre malattie polmonari rare

Da segnalare che alcune patologie possono essere incluse anche in ERN differenti, ad esempio quello delle malattie epatiche (RARE-LIVER) per il difetto di alfa1-antitripsina e quello dei tumori solidi (EURACAN) per il mesotelioma. Le malattie polmonari rare pediatriche rientrano nella maggior parte dei Core Networks (Pneumopatie Interstiziali; Fibrosi Cistica; Discinesia Ciliare; Ipertensione Polmonare; Altre Malattie Polmonari Rare), ma non in tutti.

Il Core Network "**Altre malattie polmonari rare**" è di particolare interesse per l'età pediatrica in quanto include un gruppo eterogeneo di disordini non altrimenti classificati, e cioè:
a) **Malformazioni polmonari congenite**: è un vasto spettro di condizioni la cui incidenza è stimata tra 1: 8,000 e 1: 35,000 nati vivi, spesso identificate con indagini prenatali (Eco e/o Risonanza) la cui elevata sensibilità potrebbe farne aumentare l'incidenza fino a 1: 3,000;
b) **Disordini del *drive* respiratorio**: caratterizzati da Ipoventilazione Congenita Centrale
sindrome di Ondine

sindrome di Haddad
sindrome ROHHAD (Rapid onset Obesity with Hypothalamic Dysfunction and Autonomic Dysregulation)
malformazione di Chiari ed altri difetti strutturali del cervelletto;

c) **Lesioni granulomatose e cistiche in corso di disordini genetici**:
sindrome di Birt-Hogg-Dubé (mutazioni del gene della follicolina e coinvolgimento di polmone e cute)
altri disordini, tra cui istiocitosi polmonare di Langerhans, linfangioleiomiomatosi, sindrome di Marfan,
neurofibromatosi 1, sindrome di Ehlers–Danlos;

d) **Tumori primitivi del polmone:** comprende un vasto spettro di tipi istologici tra cui
amartomi
tumori miofibroblastici infiammatori
blastomi pleuropolmonari.

Per aderire alla rete, i centri hanno dovuto certificare di seguire un minimo di pazienti in una o più categoria, di avere una produzione scientifica e/o un ruolo di leader in registri inerenti alla specifica categoria e di poter offrire una serie di *facilities* diagnostiche ed assistenziali.
In Italia, i centri che afferiscono al momento alla rete ERN Lung sono: Torino (AOU S. Luigi), Milano (Fondazione IRCCS MultiMedica S. Giuseppe), Pavia (Fondazione IRCCS S. Matteo), Trieste (Ospedali Riuniti), Padova (AO), Verona (AOU), Bologna (AOU), Modena (AOU), Forlì (Osp. Morgagni-Pierantoni), Pisa (AOU), Firenze (Ospedale Meyer), Roma (Ospedale Pediatrico Bambino Gesù), Napoli (AOU Federico II), Catania (AOU Policlinico V. Emanuele).

Nei vari core networks sono stati identificati una serie di compiti, tra cui:

7. Trials Clinici e Ricerca
8. Problemi di natura etica
9. Registri e biobanche
10. Prognosi e qualità di vita
11. Linee guida e Best Practice di assistenza
12. Training ed educazione.

GAZZETTA UFFICIALE
DELLA REPUBBLICA ITALIANA

Malattie rare esentate dalla partecipazione al costo

...ma
•Assenti malformazioni

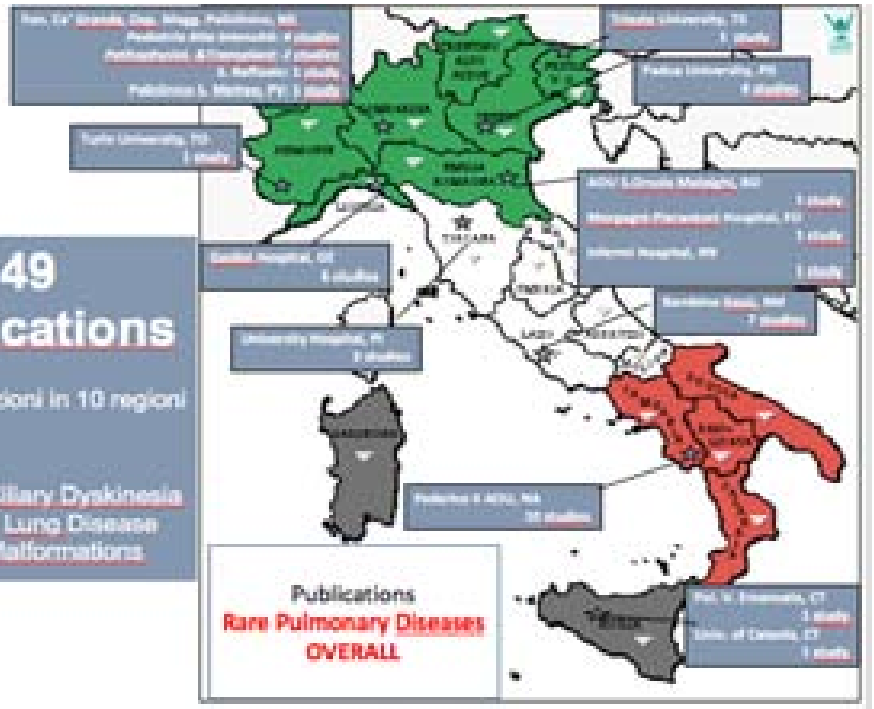
•Carenza congenita di alfa-1-antitripsina è tra le Malattie del Sistema Immunitario (!)

G.U. suppl. 12/07/2001: solo s. di Kartagener, ma tra le malformazioni!

49 publications

da 14 istituzioni in 10 regioni

1.Primary Ciliary Dyskinesia
2.Interstitial Lung Disease
3.Airways Malformations



Pubblicazioni

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Non- Cystic Fibrosis Bronchiectasis	**Patria MF**, Longhi B, Lelii M, Tagliabue C, Lavelli M, Galeone C, Principi N, **Esposito S**	Children with recurrent pneumonia and non-cystic fibrosis bronchiectasis	Ital J Pediatr;42:13.	2016	1.614	
Idiopathic Pulmonary Haemosiderosis	**Castellazzi L**, Patria MF, Frati G, Esposito AA, **Esposito S**	Idiopathic pulmonary haemosiderosis in paediatric patients: how to make an early diagnosis	Ital J Pediatr;42(1):86.	2016	1.614	
Interstitial Lung Disease	**Lelii M**, Patria MF, PinzaniR, Tenconi R, Mori A, Bonelli N**, Esposito S**	Persistent tachypnoea and intercostal retractions in a child: a case report of neuroendocrine cell hyperplasia	IJERPH International Journal of Environmental research and Public Health	2017, in press	2.101	
Oesophageal atresia with fistula	**Patria MF,** Ghislanzoni S; Macchini F; Lelii M; Mori A; Leva E; Principi N, **Esposito S**	Respiratory morbidity in children with repaired congenital oesophageal atresia with or without tracheoesophageal fistula	BMC Pulmonary Medicine	2017, in press	2.435	
Primary Ciliary Dyskinesia	**Piatti G**, De Santi MM, Torretta S, Pignataro L, Soi D, Ambrosetti U.	Cilia and Ear	Ann Otol Rhinol Laryngol.126(4):322-327.	2017		
Primary Ciliary Dyskinesia	**Piatti G,** De Santi MM, Brogi M, Castorina P, Ambrosetti U.	Emerging ciliopathies: are respiratory cilia compromised in Usher syndrome?	Am J Otolaryngol. 35(3):340-6.	2014		

Interstitial lung disease	**Carrera P,** Ferrari M, Presi S, Ventura L, Vergani B, Lucchini V, Cogo P, Carnielli V, Somaschini M, Tagliabue P	Null ABCA3 in humans: Large homozygous ABCA3 deletion, correlation to clinical-pathological findings	Pediatr Pulmonol. 49(3):E116-20.	2014	2.8	
Interstitial Lung Disease	Campo I, Zorzetto M, Mariani F, Kadija Z, Morbini P, Dore R, Kaltenborn E, Frixel S, Zarbock R, Liebisch G, Hegermann J, Wrede C, Griese M, **Luisetti M.**	A large kindred of pulmonary fibrosis associated with a novel ABCA3 gene variant.	Respir Res. 15;15:43.	2014	3.841	
Congenital Central Hypoventilation Syndrome	Biffi E, Piazza C, Cavalleri M, Taddeo P, Carcano A, Morandi F, **Reni G.**	An assistive device for outpatients during sleep.	Ann Biomed Eng. 42(10):2106-16.	2014		
Lung transplant	**Boffini M**, Venuta F, Rea F, Parisi Francesco, Marinelli G, Nanni Costa A, Rinaldi M	Urgent lung transplant programme in Italy: analysis of the first 14 months	Interact Cardiovasc Thorac Surg. 19(5):795-800; discussion 800.	2014	1,11	
Congenital Lung Malformations	Casagrande A, **Pederiva F.**	Association between Congenital Lung Malformations and Lung Tumors in Children and Adults: A Systematic Review	J Thorac Oncol. 11(11):1837-1845.	2016		
Interstitial Lung Disease	**Luca Bertelli** Salvatore Cazzato Tamara Belotti Arcangelo Prete Giampaolo Ricci **Andrea Pession**	Prevalence, Risk Factors, and Outcomes of Bronchiolitis Obliterans After Allogeneic Hematopoietic Stem Cell Transplantation	Pediatric Allergy, Immunology, and Pulmonology	2017	0,9	
Interstitial Lung Disease	Ravaglia C, Tomassetti S, Gurioli C, Piciucchi S, Dubini A, Gurioli C, Casoni GL, Romagnoli M, Carloni A, Tantalocco P, Buccioli M, Chilosi M, **Poletti V**.	Features and outcome of familial idiopathic pulmonary fibrosis.	Sarcoidosis Vasc Diffuse Lung Dis. 31(1):28-36	2014		

Idiopathic Pulmonary Haemosiderosis	Potalivo A, Finessi L, Facondini F, Lupo A, Andreoni C, Giuliani G, **Cavicchi C**.	Severe Respiratory Distress in a Child with Pulmonary Idiopathic Hemosiderosis Initially Presenting with Iron-Deficiency Anemia	Case Rep Pulmonol.;2015:876904.	2015		
Primary Ciliary Dyskinesia	**Boaretto F**, Snijders D, Salvoro C, Spalletta A, Mostacciuolo ML, Collura M, Cazzato S, Girosi D, Silvestri M, Rossi GA, Barbato A, Vazza G	Diagnosis of **Primary** Ciliary **Dyskinesia** by a Targeted Next-Generation Sequencing Panel: Molecular and Clinical Findings in Italian Patients.	J Mol Diagn;18(6):912-922	2016	4.245	Casi di Padova, Genova, Bologna e Palermo
Non- Cystic Fibrosis bronchiectasis	**Snijders D**, Fernandez Dominguez B, Calgaro S, Bertozzi I, Escribano Montaner A, Perilongo G, Barbato A	Mucociliary clearance techniques for treating non-cystic fibrosis bronchiectasis: Is there evidence?	Int J Immunopathol Pharmacol.;28(2):150-9.	2015	1.470	
Primary Ciliary Dyskinesia	**Snijders D**, Bertozzi I, **Barbato A**.	Nasal NO, high-speed video microscopy, electron microscopy, and genetics: a primary ciliary dyskinesia puzzle to complete.	Pediatr Res. 76(3):321.	2014	2.761	
Interstitial Lung Disease	**Simonato M,** Maritussio A, Pioselli B, Carnielli VP, **Cogo P**	Surfactant protein C metabolism in human infants and adult patients by stable isotope tracer and mass spectrometry	Anal Bioanal Chem. 406(25):6225-33.	2014	3,58	

Primary Ciliary Dyskinesia	**Olcese C**, Patel MP, Shoemark A, Kiviluoto S, Legendre M, Williams HJ, Vaughan CK, Hayward J, Goldenberg A, Emes RD, Munye MM, Dyer L, Cahill T, Bevillard J,Gehrig C, Guipponi M, Chantot S, Duquesnoy P, Thomas L, Jeanson L, Copin B,Tamalet A, Thauvin-Robinet C, Papon JF, Garin A, Pin I, Vera G, Aurora P, Fassad MR, Jenkins L, Boustred C, Cullup T, Dixon M, Onoufriadis A, Bush A, Chung EM, Antonarakis SE, Loebinger MR, Wilson R, Armengot M, Escudier E, Hogg C; UK10K Rare Group, Amselem S, Sun Z, Bartoloni L, Blouin JL, Mitchison HM.	X-linked primary ciliary dyskinesia due to mutations in the cytoplasmic axonemal dynein assembly factor PIH1D3.	Nat Commun. 8:14279.	2017	12	
Airways cancer	Antonescu CR, Suurmeijer AJ, Zhang L, Sung YS, Jungbluth AA, Travis WD, Al-Ahmadie H, Fletcher CD, **Alaggio R**.	Molecular characterization of inflammatory myofibroblastic tumors with frequent ALK and ROS1 gene fusions and rare novel RET rearrangement.	Am J Surg Pathol.;39(7):957-67	2015		
Airways cancer	Varela P, Pio L, **Torre M.**	Primary tracheobronchial tumors in children.	Semin Pediatr Surg. 25(3):150-5.	2016		
Airways cancer	**Sacco O,** Moscatelli A, Conte M, Grasso C, Magnano GM, Sementa AR, Martelli A, Rossi GA.	Long-Term Extracorporeal Membrane Oxygenation as Bridging Strategies to Lung Transplantation in Rapidly Devastating Isolated Langerhans Cell Histiocytosis	Pediatr Blood Cancer. 63(5):941-3.	2016		

Congenital Lung Malformations	**Sacco O**, Moscatelli A, Nozza P, Rossi GA.	Respiratory Distress in a 3-Month-Old Infant with a Mass Obstructing the Right Main-Stem Bronchus: An Unusual Localization of Infantile Hemangioma	J Pediatr. 182:397-397	2017		
Congenital Lung Malformations	**Sacco O**, Santoro F, Ribera E, Magnano GM, Rossi GA.	Short-length ligamentum arteriosum as a cause of congenital narrowing of the left main stem bronchus	Pediatr Pulmonol. 51(12):1356-1361.	2016	2.8	
Congenital Lung Malformations	Ghezzi M, Silvestri M, Sacco O, Panigada S, Girosi D, Magnano GM, **Rossi GA**	Mild tracheal compression by aberrant innominate artery and chronic dry cough in children.	Pediatr Pulmonol. 51(3):286-94.	2016	2.8	
Congenital Lung Malformations	**Mattioli G**, Pio L, Disma NM, Torre M, Sacco O, Pistorio A, Zanaboni C, Montobbio G, Barra F, Ramenghi LA.	Congenital Lung Malformations: Shifting from Open to Thoracoscopic Surgery.	Pediatr Neonatol. 57(6):463-466	2016		
Congenital Central hypoventilation syndrome	Paddeu EM, Giganti F, Piumelli R, De Masi S, Filippi L, Viggiano MP, **Donzelli G.**	Sleeping problems in mothers and fathers of patients suffering from congenital central hypoventilation syndrome.	Sleep Breath. 19(3):1057-64.	2015		
Primary Ciliary Dyskinesia	**Pifferi M**, Bush A, Caramella D, Metelli MR, Di Cicco M, Piras M, Gherarducci G, Capristo C, Maggi F, Peroni D, Boner AL.	Matrix metalloproteinases and airway remodeling and function in primary ciliary dyskinesia.	Respir Med. 124: 49-56	2017	3.217	
Primary Ciliary Dyskinesia	**Lai M**, **Pifferi M**, Bush A, Piras M, Michelucci A, Di Cicco M, del Grosso A, Quaranta P, Cursi C, Tantillo E, Franceschi S, Mazzanti MC, Simi P, Saggese G, Boner A, Pistello M.	Gene editing of DNAH11 restores normal cilia motility in primary ciliary dyskinesia.	J Med Genet. 53(4): 242-9	2016	5.451	I primi due autori hanno contribuito ugualmente al lavoro.

Primary Ciliary Dyskinesia	**Pifferi M**, Bush A, Michelucci A, Di Cicco M, Piras M, Caramella D, Mazzei F, Neri M, Pioggia G, Tartarisco G, Saggese G, Simi P, Boner AL	Mannose-binding lectin 2 gene polymorphism and lung damage in primary ciliary dyskinesia.	Pediatr Pulmonol 50(2): 179-86	2015	2.8	
Congenital Central hypoventilation syndrome	**Rossetti E**, Bianchi R, Paglietti MG, Cutrera R, Picardo S.	Severe phenotype of rapid-onset obesity, hypoventilation, hypothalamic dysfunction, and autonomic dysfunction syndrome	Minerva Anestesiol. 80(6):744-5.	2014		
Interstitial Lung Disease	**Paolini A**, Baldassarre A, Del Gaudio I, Masotti A	Structural features of the ATP-binding cassette (ABC) transporter ABCA3	Int J Mol Sci. 16(8):19631-44.	2015	2,86	
Interstitial Lung Disease	Salerno T, Peca D, Menchini L, Schiavino A, Boldrini R, Esposito F, Danhaive O, **Cutrera R.**	Surfactant Protein C-associated interstitial lung disease; three different phenotypes of the same SFTPC mutation.	Ital J Pediatr.42:23.	2016	1.614	
Interstitial Lung Disease	**Peca D**, Boldrini R, Johannson J, Shieh JT, Citti A, Petrini S, Salerno T, Cazzato S, Testa R, Messina F, Onofri A, Cenacchi G, Westermark P, Ullmann N, Cogo P, Cutrera R, Danhaive O	Clinical and ultrastructural spectrum of diffuse lung disease associated with surfactant protein C mutations	Eur J Hum Genet. 23(8):1033-41.	2015	4,35	
Interstitial Lung Disease	**Peca D**, Cutrera R, Masotti A, Boldrini R, Danhaive O	ABCA3, a key player in neonatal respiratory transition and genetic disorders of the surfactant system	Biochem Soc Trans. 43(5):913-9.	2015	3,19	
Interstitial Lung Disease	**Piersigilli F**, Peca D, Campi F, Corsello M, Landolfo F, Boldrini R, Danhaive O, Dotta A	New ATP-binding cassette A3 mutation causing surfactant metabolism dysfunction pulmonary type 3	Pediatr Int. 57(5):970-4.	2015	0,73	
Primary Ciliary Dyskinesia	Mirra V, Werner C, **Santamaria F.**	Primary Ciliary Dyskinesia: An Update on Clinical Aspects, Genetics, Diagnosis, and Future Treatment Strategies.	Front Pediatr. Jun 9;5:135.	2017		

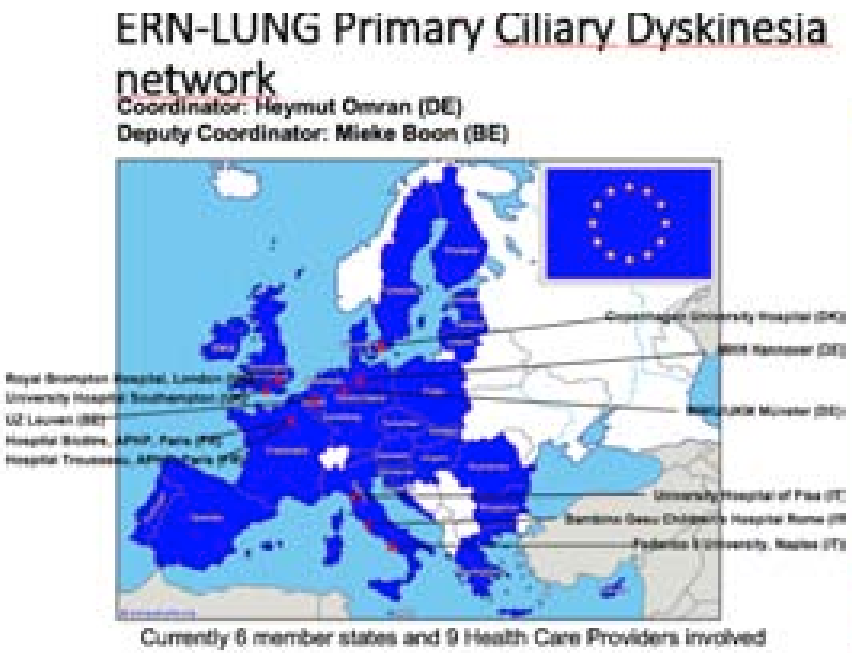
Congenital Lung Malformations	**Borgia F**, Santamaria F, Mollica C, Mongiello F, Esposito F, Palma G, Botta L, Montella S, Trimarco B, **Rapacciuolo A**	Clinical benefits, echocardiographic and MRI assessment after pulmonary sequestration treatment	Int J Cardiol, 240, 165-171	2017	6.189	
Primary Ciliary Dyskinesia	**Maglione M***, **Montella S***, Mollica C, Carnovale V, Iacotucci P, De Gregorio F, Tosco A, Cervasio M, Raia V, **Santamaria F**	Lung structure and function similarities between primary ciliary dyskinesia and mild cystic fibrosis: a pilot study	Ital J Pediatr, 43, 34	2017	1.614	
Oesophageal atresia with fistula	**Mirra V**, Maglione M, Di Micco LL, Montella S, **Santamaria F**	Longitudinal Follow-up of Chronic Pulmonary Manifestations in Esophageal Atresia: A Clinical Algorithm and Review of the Literature	Pediatr Neonatol, 58, 8-15	2017	1.287	
Interstitial Lung Disease	**Montella S**, Vece TJ, Langston C, Carrera P, Nogee LM, Hamvas A, Manna A, Cervasio M, **Santamaria F**	A disorder of surfactant metabolism without identified genetic mutations	Ital J Pediatr, 41, 93	2015	1.614	
Primary Ciliary Dyskinesia	**Mirra V**, Caffarelli C, Maglione M, Valentino R, Perruolo G, Mazzarella C, Di Micco LL, Montella S, **Santamaria F**	Hypovitaminosis D: a novel finding in primary ciliary dyskinesia	Ital J Pediatr, 41, 14	2015	1.614	
Primary Ciliary Dyskinesia	**Maglione M**, Montella S, Mirra V, Bruzzese D, **Santamaria F**	Long-term assessment of quality of life in primary ciliary dyskinesia: time for new tools?	Chest, 146, e232-e233	2014	6.044	
Primary Ciliary Dyskinesia	**Manna A**, Montella S, Maniscalco M, Maglione M, **Santamaria F**	Clinical application of nasal nitric oxide measurement in pediatric airway diseases	Pediatr Pulmonol, 50, 85-99	2015	2.8	
Primary Ciliary Dyskinesia	**Montuschi P**, Paris D, Montella S, Melck D, Mirra V, Santini G, Mores N, Montemitro E, Majo F,	Nuclear magnetic resonance-based metabolomics discriminates primary	Am J Respir Crit Care Med, 190, 229-33	2014	13	

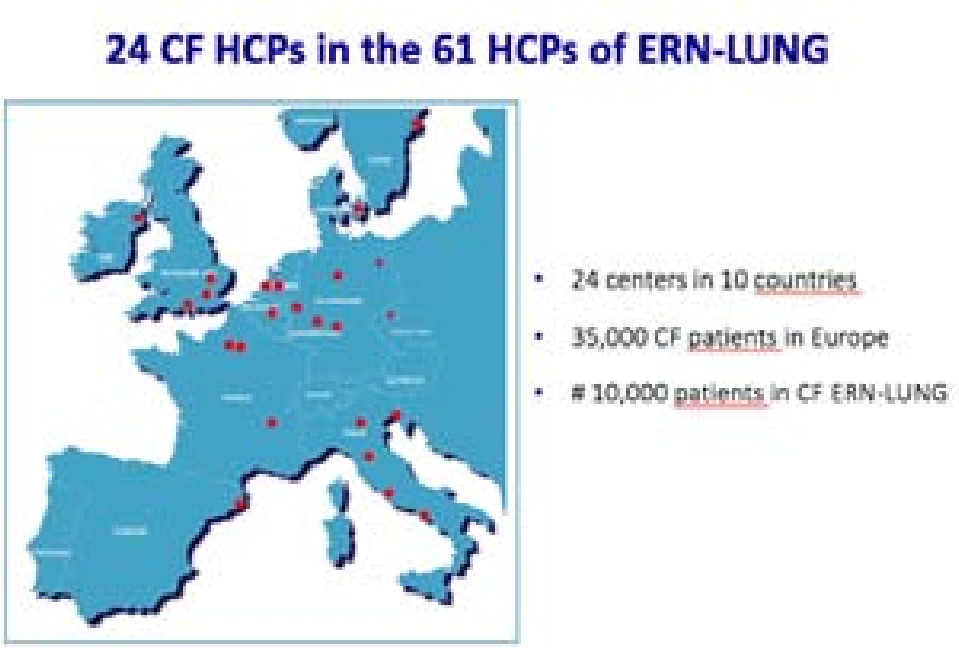
	Lucidi V, Bush A, Motta A, **Santamaria F**	ciliary dyskinesia from cystic fibrosis				
Primary Ciliary Dyskinesia	**Santamaria F**, Esposito M, Montella S, Cantone E, Mollica C, De Stefano S, Mirra V, **Carotenuto M**	Sleep disordered breathing and airway disease in primary ciliary dyskinesia	Respirology, 19, 570-5	2014	3.256	
Primary Ciliary Dyskinesia	**Maglione M**, Bush A, Nielsen KG, Hogg C, Montella S, Marthin JK, Di Giorgio A, **Santamaria F**	Multicenter analysis of body mass index, lung function, and sputum microbiology in primary ciliary dyskinesia	Pediatr Pulmonol, 49, 1243-50	2014	2.8	

Trial clinici farmacologici

1 **Nome malattia rara**	2 ***Sponsor***	3 ***Partners***		4 **Ruolo**		5 **Denominazione del *trial***	6 **Farmaco**	7 **Durata**		8 **Note**
		I	**N**	**C**	**P**			**C**	**T (anno)**	
Discinesia Ciliare Primaria (PCD)	Parion Sciences	X		X		A Phase II Randomized, Double-blind, Placebo-controlled, Incomplete Block Crossover Study to evaluate the safety and efficacy of VX-371 Solution for Inhalation in subjects with Primary Ciliary Dyskinesia	VX-371	X		Università Pisa
Interstitial Lung Disease		x			x	Hydroxychloroquine in ILD	Hydroxychloroquine	C		Università Padova
Interstitial Lung Disease		x			x	European Management Platform for Childhood Interstitial Lung Diseases - chILD-EU Register and Biobank (chILD-EU)		C		Università Padova
Interstitial Lung Disease		x			x	Hydroxychloroquine in ILD	Hydroxychloroquine	C		Osp Bambino Gesù, Roma

Malattie Respiratorie Infantili e Partecipazione Nazionale a ERN





Malattie endocrinologiche pediatriche

A cura della Società Italiana di Endocrinologia e Diabetologia Pediatrica – SIEDP
(Presidente: *Franco Cerutti*)

Antonio Balsamo
Marco Cappa
Graziano Grugni

Pubblicazioni e *trial* clinici farmacologici

Malattie endocrinologiche pediatriche

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Si stima che circa 1/3 delle malattie rare del bambino e dell'adolescente siano seguite presso i Centri con pediatri soci della Società Italiana di Endocrinologia e Diabetologia Pediatrica (SIEDP), sia perché di stretta pertinenza endocrinologica e diabetologica, sia per le complicanze di questa natura a carico di altri quadri patologici. A partire da queste premesse, la SIEDP ha avviato tra la fine del 2015 ed i primi mesi del 2016 un'indagine conoscitiva preliminare, attraverso il coinvolgimento dei Referenti dei Centri afferenti al Network italiano di Endocrinologia e Diabetologia Pediatrica, mirata a conoscere il numero di pazienti con malattia rara ivi osservati, così da poter classificare quelle più comunemente seguite. Tra gli oltre 9000 pazienti censiti, le principali patologie rilevate sono state, in ordine decrescente di numero, l'Ipotiroidismo congenito permanente, l'Ipopituitarismo, la Pubertà precoce, la Sindrome adreno-genitale, la Sindrome di Turner, le Obesità genetiche, la Disormogenesi tiroidea, il MODY e la Sindrome di Noonan. Su queste basi la nostra Società si è attivata per la stesura dei PDTA delle malattie rare e/o croniche di propria pertinenza, attraverso la costituzione di un apposito Gruppo di Lavoro, con lo scopo di produrre documenti validati su scala nazionale anziché parcellizzata in molteplici approcci localistici. Ha inoltre elaborato un piano formativo sull'argomento per i nostri Soci, e non solo, mirato ad organizzare Corsi residenziali ed eventi FAD sull'argomento specifico delle malattie rare (Progetto FOR.MA.RE: FORmazione MAlattie raRE). Questo specifico interesse viene confermato dai dati relativi alle pubblicazioni, ai trial clinici farmacologici ed alla partecipazione ad Endo-ERN.

Pubblicazioni e Trial clinici farmacologici. Il censimento delle pubblicazioni dei Soci sulle malattie rare è stato effettuato attraverso la ricerca su PubMed, relativamente al periodo gennaio 2014 – giugno 2017, considerando quelle in cui il coordinatore del lavoro risulta appartenente ad un'istituzione italiana. Si tratta complessivamente di 380 lavori, di cui 128 sono del 2014, 124 del 2015, 97 del 2016 e 31 del 2017, con una produzione media di 108 all'anno. Da una prima analisi dei dati emerge che la metà degli articoli è frutto della collaborazione di due o più gruppi di studio. Le riviste su cui sono comparsi i lavori sono in tutto 150, di cui le prime quattro per numero di pubblicazioni sono: Hormone Research in Paediatrics (n. 29; IF attuale: 1.661), Italian Journal of Pediatrics (n. 23; IF attuale: 1.614), Journal of Clinical Endocrinology and Metabolism (n. 18; IF attuale: 5.531) e American Journal of Medical Genetics (n. 18; IF attuale: 2,082). L'IF totale è 966.1, con una media di 2.549 per pubblicazione, mentre il numero maggiore di lavori è comparso su riviste con un IF compreso tra 0.1 e 5 (n. 296). In relazione ai Trial clinici farmacologici, quelli finora pervenuti hanno riguardato il deficit di ormone di crescita e la sindrome di Prader-Willi.

Pubblicazioni

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Chromosomal disorder	**Baldassarre G**, Mussa A, Banaudi E, Rossi C, Tartaglia M, Silengo M, Ferrero GB.	Phenotypic variability associated with the invariant SHOC2 c.4A>G (p.Ser2Gly) missense mutation.	Am J Med Genet A 164A(12):3120-5.	2014	2,159	
RASopathies	**Alfieri P**, Piccini G, Caciolo C, Perrino F, Gambardella ML, Mallardi M, Cesarini L, Leoni C, Leone D, Fossati C, Selicorni A, Digilio MC, Tartaglia M, Mercuri E, Zampino G, Vicari S.	Behavioral profile in RASopathies.	Am J Med Genet A 164A(4):934-42.	2014	2,159	
Holt-Oram syndrome	**Baban A**, Pitto L, Pulignani S, Cresci M, Mariani L, Gambacciani C, Digilio MC, Pongiglione G, Albanese S.	Holt-Oram syndrome with intermediate atrioventricular canal defect, and aortic coarctation: functional characterization of a de novo TBX5 mutation.	Am J Med Genet A 164A(6):1419-24.	2014	2,159	
Rubinstein-Taybi syndrome	**Marzuillo P**, Grandone A, Luongo C, Cantelmi G, Polito C, del Giudice EM, Perrone L.	Brain magnetic resonance in the routine management of Rubinstein-Taybi syndrome (RTS) can prevent life-threatening events and neurological deficits.	Am J Med Genet A 164A(8):2129-32.	2014	2,159	
Septo-optic dysplasia.	**Severino M**, Allegri AE, Pistorio A, Roviglione B, Di Iorgi N, Maghnie M, Rossi A.	Midbrain-hindbrain involvement in septo-optic dysplasia.	Am J Neuroradiol 35(8):1586-92.	2014	3,124	
Prader-Willi syndrome	**Bedogni G**, Grugni G, Tringali G, Agosti F, Sartorio A.	Assessment of fat-free mass from bioelectrical impedance analysis in obese women with Prader-Willi syndrome.	Ann Hum Biol26:1-5.	2014	1,273	

Chromosomal disorder	**Stagi S**, Lapi E, Seminara S, Guarducci S, Pantaleo M, Giglio S, Chiarelli F, de Martino M.	Long-term auxological and endocrinological evaluation of patients with 9p trisomy: a focus on the growth hormone-insulin-like growth factor-I axis.	BMC Endocr Disord 14:3.	2014	1,710	
Precocious puberty	**Lucaccioni L**, Schwahn BC, Donaldson M, Giacomozzi C.	Central precocious puberty in a 3 year-old girl with Phenylketonuria: a rare association?	BMC Endocr Disord 14:38.	2014	1,710	
Congenital hypothyroidism	**Vincenzi M**, Camilot M, Ferrarini E, Teofoli F, Venturi G, Gaudino R, Cavarzere P, De Marco G, Agretti P, Dimida A, Tonacchera M, Boner A, Antoniazzi F.	Identification of a novel pax8 gene sequence variant in four members of the same family: from congenital hypothyroidism with thyroid hypoplasia to mild subclinical hypothyroidism.	BMC Endocr Disord 14:69.	2014	1,710	
22q11.2 deletion syndrome	**Cirillo E**, Giardino G, Gallo V, Puliafito P, Azzari C, Bacchetta R, Cardinale F, Cicalese MP, Consolini R, Martino S, Martire B, Molinatto C, Plebani A, Scarano G, Soresina A, Cancrini C, Rossi P, Digilio MC, Pignata C.	Intergenerational and intrafamilial phenotypic variability in 22q11.2 deletion syndrome subjects.	BMC Med Genet 15:1.	2014	2,083	
Noonan syndrome	**Lepri FR**, Scavelli R, Digilio MC, Gnazzo M, Grotta S, Dentici ML, Pisaneschi E, Sirleto P, Capolino R, Baban A, Russo S, Franchin T, Angioni A, Dallapiccola B.	Diagnosis of Noonan syndrome and related disorders using target next generation sequencing.	BMC Med Genet 15:14.	2014	2,083	
1p36 deletion syndrome	**Stagi S**, Lapi E, Pantaleo M, Chiarelli F, Seminara S, de Martino M.	Type II diabetes and impaired glucose tolerance due to severe hyperinsulinism in patients with 1p36 deletion syndrome and a Prader-Willi-like phenotype.	BMC Med Genet 15:16.	2014	2,083	

Williams-Beuren syndrome	**Stagi S**, Lapi E, D'Avanzo MG, Perferi G, Romano S, Giglio S, Ricci S, Azzari C, Chiarelli F, Seminara S, de Martino M.	Coeliac disease and risk for other autoimmune diseases in patients with Williams-Beuren syndrome.	BMC Med Genet 15:61.	2014	2,083	
SHOX disorder	**Gatta V**, Palka C, Chiavaroli V, Franchi S, Cannataro G, Savastano M, Cotroneo AR, Chiarelli F, Mohn A, Stuppia L.	Spectrum of phenotypic anomalies in four families with deletion of the SHOX enhancer region.	BMC Med Genet 15:87.	2014	2,083	
Wolfram syndrome	**Mozzillo E**, Delvecchio M, Carella M, Grandone E, Palumbo P, Salina A, Aloi C, Buono P, Izzo A, D'Annunzio G, Vecchione G, Orrico A, Genesio R, Simonelli F, Franzese A.	A novel CISD2 intragenic deletion, optic neuropathy and platelet aggregation defect in Wolfram syndrome type 2.	BMC Med Genet 15:88.	2014	2,083	
4A syndrome	**Aragona P**, Rania L, Roszkowska AM, Puzzolo D, Micali A, Pisani A, Salzano G, Messina MF.	4A syndrome: ocular surface investigation in an Italian young patient.	BMC Ophthalmol 14:155.	2014	1,020	
Autoimmune polyendocrine syndrome	**Improda N**, Capalbo D, Cirillo E, Cerbone M, Esposito A, Pignata C, Salerno M.	Cutaneous vasculitis in patients with autoimmune polyendocrine syndrome type 1: report of a case and brief review of the literature.	BMC Pediatr 14:272.	2014	1,930	
Allan-Herndon-Dudley syndrome	**Azzolini S**, Nosadini M, Balzarin M, Sartori S, Suppiej A, Mardari R, Greggio NA, Toldo I.	Delayed myelination is not a constant feature of Allan-Herndon-Dudley syndrome: report of a new case and review of the literature.	Brain Dev 36(8):716-20.	2014	1,542	
Disorder of sexual development	**Balsamo A**, Baronio F, Berra M, Bertelloni S, D'Alberton F, Marrocco G, Vallasciani S.	Comment on "complete androgen insensitivity syndrome: optimizing diagnosis and management".	Case Rep Obstet Gynecol 2014:285715.	2014	0,000	

Kasabach-merritt syndrome	**Nakib G**, Calcaterra V, Quaretti P, Moramarco LP, Bonalumi G, Brunero M, Pelizzo G.	Chemotherapy and surgical approach with repeated endovascular embolizations: safe interdisciplinary treatment for kasabach-merritt syndrome in a small baby.	Case Rep Oncol 7(1):23-8.	2014	0,000	
Stuve-Wiedemann syndrome	**Buonuomo PS**, Macchiaiolo M, Cambiaso P, Rana I, Digilio MC, Bartuli A.	Long-term follow-up in Stuve-Wiedemann syndrome: a case report with articular involvement.	Clin Dysmorphol 23(2):45-6.	2014	0,608	
GH deficiency	**Capalbo D**, Mattace Raso G, Esposito A, Di Mase R, Barbieri F, Meli R, Bruzzese D, Salerno M.	Cluster of cardiometabolic risk factors in children with GH deficiency: a prospective, case-control study.	Clin Endocrinol (Oxf) 80(6):856-62.	2014	3,457	
Prader-Willi syndrome	**Rigamonti AE**, Bini S, Grugni G, Agosti F, De Col A, Mallone M, Cella SG, Sartorio A.	Unexpectedly increased anorexigenic postprandial responses of PYY and GLP-1 to fast ice cream consumption in adult patients with Prader-Willi syndrome.	Clin Endocrinol (Oxf) 81(4):542-50.	2014	3,457	
Congenital hyperinsulinism	**Maiorana A**, Barbetti F, Boiani A, Rufini V, Pizzoferro M, Francalanci P, Faletra F, Nichols CG, Grimaldi C, de Ville de Goyet J, Rahier J, Henquin JC, Dionisi-Vici C.	Focal congenital hyperinsulinism managed by medical treatment: a diagnostic algorithm based on molecular genetic screening.	Clin Endocrinol (Oxf) 81(5):679-88.	2014	3,457	
Adrenal disorders	**Dalla Costa M**, Bonanni G, Masiero S, Faggian D, Chen S, Furmaniak J, Rees Smith B, Perniola R, Radetti G, Garelli S, Chiarelli S, Albergoni MP, Plebani M, Betterle C.	Gonadal function in males with autoimmune Addison's disease and autoantibodies to steroidogenic enzymes.	Clin Exp Immunol 176(3):373-9.	2014	3,037	
Haematologic disorders, general	**Rolla R**, Castagno M, Zaffaroni M, Grigollo B, Colombo S, Piccotti S, Dellora C, Bona G, Bellomo G.	Neonatal screening for sickle cell disease and other hemoglobinopathies in "the changing Europe".	Clin Lab 60(12):2089-93.	2014	1,129	

Prader-Willi syndrome	**Cimolin V**, Galli M, Rigoldi C, Grugni G, Vismara L, de Souza SA, Mainardi L, Albertini G, Capodaglio P.	The fractal dimension approach in posture: a comparison between Down and Prader-Willi syndrome patients.	Comput Methods Biomech Biomed Engin 17(14):1535-41.	2014	1,793	
Diabetes mellitus	**Bonura C**, Frontino G, Rigamonti A, Battaglino R, Favalli V, Ferro G, Rubino C, Del Barba P, Pesapane F, Nazzaro G, Gianotti R, Bonfanti R, Meschi F, Chiumello G.	Necrobiosis Lipoidica Diabeticorum: A pediatric case report.	Dermatoendocrinol 6(1):e27790.	2014	0,000	
MODY	**Delvecchio M**, Ludovico O, Menzaghi C, Di Paola R, Zelante L, Marucci A, Grasso V, Trischitta V, Carella M, Barbetti F, Gallo F, Coccioli MS, Zecchino C, Faienza MF, Cardinale G, Franzese A, Mozzillo E, Iafusco D, Zanfardino A.	Low prevalence of HNF1A mutations after molecular screening of multiple MODY genes in 58 Italian families recruited in the pediatric or adult diabetes clinic from a single Italian hospital.	Diabetes Care 37(12):e258-60.	2014	8,420	
Prader-Willi syndrome	**Fintini D**, Grugni G, Brufani C, Bocchini S, Cappa M, Crinò A.	Use of GLP-1 receptor agonists in Prader-Willi Syndrome: report of six cases.	Diabetes Care 37(4):e76-7.	2014	8,420	
Neonatal diabetes mellitus	**Iafusco D**, Salardi S, Chiari G, Toni S, Rabbone I, Pesavento R, Pasquino B, de Benedictis A, Maltoni G, Colombo C, Russo L, Massa O, Sudano M, Cadario F, Porta M, Barbetti F; Early Onset Diabetes Study Group of the Italian Society of Pediatric Endocrinology and Diabetology (ISPED)	No sign of proliferative retinopathy in 15 patients with permanent neonatal diabetes with a median diabetes duration of 24 years.	Diabetes Care 37(8):e181-2.	2014	8,420	

Neonatal diabetes mellitus	**Passanisi S**, Timpanaro T, Lo Presti D, Mammì C, Caruso-Nicoletti M.	Treatment of transient neonatal diabetes mellitus: insulin pump or insulin glargine? Our experience.	Diabetes Technol Ther 16(12):880-4.	2014	2,106	
Disorder of sexual development	**Laino L**, Majore S, Preziosi N, Grammatico B, De Bernardo C, Scommegna S, Rapone AM, Marrocco G, Bottillo I, Grammatico P.	Disorders of sex development: a genetic study of patients in a multidisciplinary clinic.	Endocr Connect 3(4):180-92.	2014	0,000	
Rare diseases (review)	**Polizzi A**, Balsamo A, Bal MO, Taruscio D.	Rare diseases research and practice.	Endocr Dev 27:234-56.	2014	4,022	
Adrenal disorders	**Guaraldi F**, Parasiliti-Caprino M, Goggi R, Beccuti G, Grottoli S, Arvat E, Ghizzoni L, Ghigo E, Giordano R, Gori D.	Identification of risk conditions for the development of adrenal disorders: how optimized PubMed search strategies makes the difference.	Endocrine 47(3):734-9.	2014	1,997	
GH deficiency	**Babu D**, Mellone S, Fusco I, Petri A, Walker GE, Bellone S, Prodam F, Momigliano-Richiardi P, Bona G, Giordano M.	Novel mutations in the GH gene (GH1) uncover putative splicing regulatory elements.	Endocrinology 155(5):1786-92.	2014	6,209	
Chromosomal disorder	**Surace C**, Berardinelli F, Masotti A, Roberti MC, Da Sacco L, D'Elia G, Sirleto P, Digilio MC, Cusmai R, Grotta S, Petrocchi S, El Hachem M, Pisaneschi E, Ciocca L, Russo S, Lepri FR, Sgura A, Angioni A.	Telomere shortening and telomere position effect in mild ring 17 syndrome.	Epigenetics Chromatin 7(1):1.	2014	5,333	
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Autoimmune polyendocrine syndrome	**Valenzise M**, Aversa T, Salzano G, Zirilli G, De Luca F, Su M.	Novel insight into Chronic Inflammatory Demyelinating Polineuropathy in APECED syndrome: molecular mechanisms and clinical implications in children.	Ital J Pediatr 43:11.	2017	1,614	

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Prader-Willi syndrome	**Mele C**, Grugni G, Mai S, Vietti R, Aimaretti G, Scacchi M, Marzullo P	Circulating angiopoietin-like 8 (ANGPTL8) is a marker of liver steatosis and is negatively regulated by Prader-Willi Syndrome	Sci Rep. 2017 7: 3186; DOI:10.1038/s41598-017-03538-7	2017	5,228	
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Trial clinici farmacologici

1 **Nome MR**	2 ***Sponsor***	3 ***Partners***		4 **Ruolo**		5 **Denominazione del *trial***	6 **Farmaco**	7 **Durata**		8 **Note**
		I	**N**	**C**	**P**			**C**	**T (anno)**	
Iposodiemia cronica (ipoaldosteronismo, Pseudoipoaldosteronismo e altre cause secondarie	OTZUKA	**X**			**X**	Studio randomizzato multicentrico in aperto di fase 3b sugli effetti della sospensione di SAMSCAm (Tolvaptan) titolato sul sodio	156-08-276 (Tolvactan)	**X**	**2018**	UOC di Endocrinologia, DPUO, Ospedale Pediatrico Bambino Gesù,IRCCS, Roma
Sindrome di Laron	IPSEN	**X**			**X**	European Increlex^{Ò} (mecasermin [rDNA origin] injection) growth forum database:	2 79 52800 002 (Increlex)	**X**		UOC di Endocrinologia, DPUO, Ospedale Pediatrico Bambino Gesù,IRCCS, Roma
Sindrome di Allan-Herndon-Dudley	Un. Rotterdam	**X**			**X**	Trattamento con analogo ormone T3 (TRIAC) in pazienti con mutazione dell'MTC8	MCT-8-2014-1	**X**	**2018**	UOC di Endocrinologia, DPUO, Ospedale Pediatrico Bambino Gesù,IRCCS, Roma
X-linked Adrenoleucodistrofia	Pharmaelle	**X**		**X**		Approccio nutrizionale innovativo al trattamento dell'adrenoleucodistrofia X - Linked	ALDIXYL	**X**		UOC di Endocrinologia, DPUO, Ospedale Pediatrico Bambino Gesù,IRCCS, Roma

Sindrome di Prader-Willi	Alizè Pharma	X			X	A Phase IIa, randomized, double-blind, placebo-controlled, multi-center study to evaluate the safety, tolerability, and effects of AZP-531, an Unacylated Ghrelin analog, on food-related behavior in patients with Prader-Willi Syndrome	AZP-531		X (2016)	1) Divisione di Auxologia - IRCCS Ospedale S. Giuseppe di Piancavallo, Istituto Auxologico Italiano, Verbania (VB) 2) Struttura Semplice di Patologia Endocrina Autoimmune – UOC di Endocrinologia e Diabetologia - Ospedale Pediatrico Bambino Gesù, IRCCS Palidoro (Roma)
T1D	Horizon 2020	X				INNODIA	IL-2	X		Cattedra di Pediatria, Università di Chieti
Deficit GH	OPKO	X				GH Treatment	Long Acting GH	X		Cattedra di Pediatria, Università di Chieti

Partecipazione Nazionale a Endo-ERN

[1]Graziano Grugni, [2]Marco Cappa, [3]Antonio Balsamo

[1]Divisione di AuxologiaIRCCS Ospedale S. Giuseppe Istituto Auxologico ItalianoPiancavallo di Oggebbio (VB)

[2]Head of Endocrinology and Diabetic Unit Bambino Gesù Children's Hospital, IRCCS Rome

[3] AOU Sant'Orsola Malpighi, Department of Medical and Surgical Sciences; Pediatric Unit - Endocrinology Program, Bologna

Il 1° bando per la partecipazione agli European Reference Networks (ERNs) per le malattie rare è stato lanciato nel marzo del 2016. La prima Assemblea Generale (GA) della Rete di Riferimento Europea sulle Condizioni Endocrine Rare (Endo-ERN) si è svolta il 27 marzo 2017 a Leiden, in Olanda. Settanta dei 71 Coordinatori che rappresentano i Centri Europei delle Strutture Ospedaliero-Universitarie (HCPs: Health Care Providers) attualmente parte di Endo-ERN, hanno partecipato all'assemblea. Dieci di questi Centri rappresentavano HCPs Italiani, con sede a Bologna (AOU S.Orsola-Malpighi), Firenze (2; AOU Careggi; Ospedale Meyer), Genova (IRCCS S.Martino), Milano (2; IRCCS Auxologico; Ospedale S.Raffaele), Napoli AOU Federico II), Padova (AOU Padova), Pisa (AOU Pisana), Torino (AOU Città della salute e della Scienza). Nel corso dell'assemblea è emerso il ruolo importante rivestito dall'Italia, i cui Centri sono presenti complessivamente in tutte le otto aree tematiche della rete. Dei 10 HCP presenti, 4 sono stati rappresentati ufficialmente da un coordinatore di area pediatrica (Bologna-S.Orsola-Malpighi, Firenze-Meyer, Milano-S.Raffaele, Padova-A.O.U.- Pediatria). L'Italia con l'Olanda e la Francia si sono organizzati con un coordinatore nazionale (Annamaria Colao per l'area Adulti e Marco Cappa per l'area pediatrica).

Un "Centro di riferimento" per le malattie rare deve prevedere un Gruppo coordinato multidisciplinare con personale sanitario esperto e competente per il trattamento di queste condizioni. Il Centro deve avere al suo interno la possibilità di assistere le persone dall'età pediatrica fino all'età adulta, dedicarsi ad attività di ricerca e provvedere alla formazione ed aggiornamento dei professionisti che lavorano al suo interno. Caratteristica innovativa e caratterizzante è il coinvolgimento di rappresentanti di "Pazienti" e Gruppi di Supporto. Endo-ERN ha attivato 8 Principali Gruppi Tematici (MTGs: Main Thematic Groups), che riguardano tutto lo spettro delle condizioni endocrinologiche rare o ad esse correlabili: Surrene (MTG1), Alterazioni dell'equilibrio calcio-fosforo (MTG2), Alterazioni genetiche dell'equilibrio glucosio-insulina (MTG3), Sindromi da tumori endocrini genetici (MTG4), Crescita e sindromi con obesità genetiche (MTG5), Ipofisi (MTG6), Sviluppo e maturazione sessuale (MTG7), Tiroide (MTG8). In ognuno degli 8 MTG sono stati individuati alcuni sottotemi di interesse (es. MTG6 Ipofisi: sottotemi: 1) adenoma ipofisario; 2) ipopituitarismo congenito e 3) ipopituitarismo acquisito) e 5 "pacchetti di lavoro" (WPs: Work Packages) che riguardano: Istruzione & Insegnamento (WP1); Tele-Medicina & Interfacce di comunicazione/interazione (WP2); Ricerca & Scienza (WP3); Qualità dell'assistenza & punto di vista del paziente (WP4); Diagnostica & indagini di Laboratorio (WP5). Ogni Centro accreditato per lo specifico MTG, può scegliere a quale sottotema e WP partecipare attivamente.

In base ad una indagine conoscitiva preliminare della SIEDP, la totalità dei Centri di riferimento endocrinologici pediatrici accreditati

dal Ministero Italiano della Salute appartengano ad HCPs rappresentate da soci SIEDP. La fotografia attuale dei centri Nazionali inclusi nella rete Europea Endo-ERN, con le rispettive aree tematiche richieste/accreditate e i pacchetti di lavoro (WP) opzionati dai singoli Centri (quando segnalati), sono riportate nella Tabella:

Tabella:

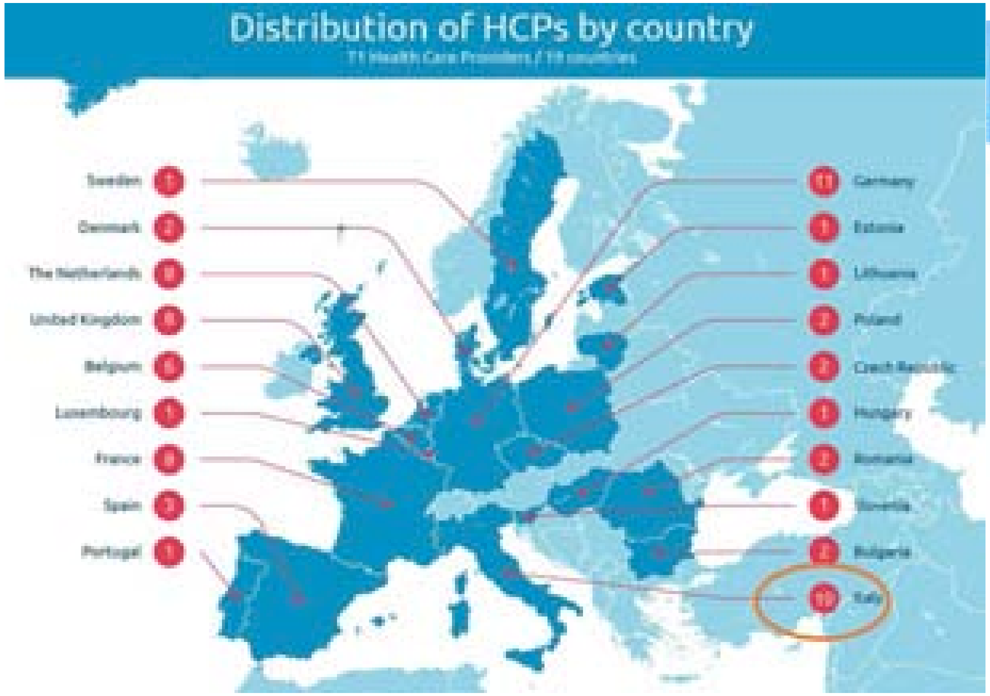
HCPs	Ambito	MTG 1	MTG 2	MTG3	MTG4	MTG5	MTG6	MTG7	MTG8
AOU Bologna	P (UO Ped)	SI (WP1-5)	NR	SI (WP nd)	SI (WP nd)	SI (WP 1,4)	SI (WP1-5)	SI (WP 1-5)	SI (WP1,4)
S.Orsola-Malpighi	A (UO Endo)	SI (WP1-5)	NR	SI (WP nd)	SI (WP nd)	SI (WP 1-5)	SI (WP1-5)	SI (WP 1-5)	SI (no WP)
AOU Firenze	P (Meyer)	NR	NR	SI (WP1,5)	NR	NR	NR	NR	NR
Careggi	A (UO Endo)	NR	SI (WP nd)	NR	SI (WP nd)	NR	NR	SI (WP nd)	NR
AOU Genova	P (Gaslini)	SI Endorsement Nazionale			2a call Europea ERN				
IRCCS S.Martino	A (S.Martino)	NR	NR	NR	NR	NR	SI (WP1-5)	NR	NR
AOU Milano	P (HS.Raffaele)	SI (WP nd)	SI (WP nd)	NR	SI (WP nd)	SI (WP nd)	SI (WP nd)	SI (WP1-5)	SI (WP nd)
	A (I. Auxologico)	SI (WP nd)	SI (WP nd)	NR	SI (WP 1-5)	SI (WP1-5)	SI (WP1-5)	SI (WP1-5)	SI (WP nd)
AOU Napoli	P (UO Ped)	SI (WP nd)	SI (WP nd)	SI (WP nd)	SI (WP nd)	SI (WP nd)	SI (WP nd)	SI (WP nd)	SI (WP nd)
Federico II	A (UO Endo)	SI (WP nd)	SI (WP nd)	SI (WP nd)	SI (WP nd)	SI (WP nd)	SI (WP nd)	SI (WP nd)	SI (WP nd)
AOU Padova	P (US Ped)	SI (WP1-5)	SI (WP nd)	NR	SI (WP1-5)	SI (WP1-5)	SI WP1-5)	SI (WP 1-5)	SI (WP 1,2,5)
	A (UO Endo)	SI (WP1-5)	SI (WP nd)	SI (WP nd)	SI (WP1-5)	SI (WP1-5)	SI (WP1-5)	SI (WP 1-5)	SI (WP 1,2,5)
AOU Pisa	P (UO Ped)	2a call Europea ERN							
	A (UO Endo)	NR	NR	SI (WP1,3)	NR	NR	NR	NR	NR
AOU ROMA	P (Bambin Gesù)	SI Endorsement Nazionale			2a call Europea ERN				
	A (Sapienza UO En)	2a call Europea ERN							
AOU Torino	P (R. Margherita)	SI Endorsement Nazionale			2a call Europea ERN				
Città Sal.& Sc.	A (UO Endo)	SI (WP1-4)	NR	SI (WP nd)	SI (WG nd)	NR	SI (WP1-4)	NR	SI(WP 5)

P: Endocrinologia Pediatrica; A: Endocrinologia Adulto; SI: accettato; NR: Non Richiesto; nd: non definiti/comunicati;

Secondo quanto dichiarato dal coordinatore dell'ENDO-ERN è verosimile che entro il 2017 sarà possibile riaprire il Bando per i centri che hanno ricevuto l'endorsement ministeriale ma ancora non figurano ufficialmente della rete.

Vi è grande attenzione per i registri di patologia e vi è la prospettiva di sviluppare registri Europei più avanzati e condivisibili.

Auspichiamo che il contributo della nostra Società, insieme a tutte le altre con le quali vi sono interessi comuni, possa contribuire al successo di questa sfida sanitaria Europea. Tutte le informazioni e la documentazione relativa all'iter di approvazione di ENDO-ERN sono disponibili sul sito www.endo-ern.eu.



Italy	Azienda Ospedaliera Universitaria di Padova	Scaroni Greggio	Carla Nella	HCP representatives
Italy	Azienda Ospedaliera Universitaria Sant' Orsola Malpighi, Bologna	Balsamo Gambineri	Antonio Alessandra	HCP representatives
Italy	Azienda Ospedaliera Universitaria "Federico II", Napoli	Colao * Salerno	Annamaria Mariacarolina	HCP representatives
Italy	Azienda Ospedaliera Universitaria Pisana	[illegible]	Giovanni	HCP representative
Italy	Azienda Ospedaliera Universitaria Città della Salute e della Scienza di Torino	Giordano	Roberta	HCP representative
Italy	IRCCS A.O.U San Martino, Genova	Ferone	Diego	HCP representative
Italy	IRCCS Istituto Auxologico Italiano – Milano	Persani **	Luca	HCP representative
Italy	Ospedale San Raffaele – Milano	Russo	Gianni	HCP representative
Italy	Azienda Ospedaliera Universitaria Careggi - Firenze	Brandi	Maria Luisa	HCP representative
Italy	Meyer Children's Hospital – Firenze	[illegible]	Barbara	HCP representative
Italy	European Patient Advocacy Groups	Vitali	Diana	ePAG representative

* Advisory Board member
** MTG chair
Colao AM and Cappa M: National Coordinators for Adult and Pediatric Endocrinology, respectively

Malattie metaboliche ereditarie

A cura della Società Italiana per lo studio delle malattie Metaboliche Ereditarie e lo Screening Neonatale - SIMMESN
(Presidente: *Carlo Dionisi Vici*)

Daniela Concolino
Carlo Dionisi Vici
Francesco Porta

Pubblicazioni

Attività di ricerca relative alle malattie rare pediatriche, svolte da centri italiani, nel settore specialistico Malattie Metaboliche, SIMMESN

Daniela Concolino
Università Magna Graecia di Catanzaro, Centro Pediatria Genetica e Malattie rare

La comunità scientifica della SIMMESN - composta da pediatri, biologi, biochimici, dietisti, psicologi, ecc. - è costituita da 170 soci afferenti a 40 Centri clinici o laboratoristici distribuiti su tutto il territorio nazionale. Per il forum SIRP è stata analizzata l'attività di ricerca negli anni 2014-17, in cui il socio SIMMESN ha svolto un ruolo leader nei seguenti 3 ambiti:

PRODUZIONE SCIENTIFICA

Nel periodo gennaio 2014 - giugno 2017 sono stati prodotti 189 articoli, sia di ricerca clinica che di ricerca di base. Gli articoli sono stati censiti tramite PubMed e sono stati analizzati solo quelli dove il socio è risultato essere primo/ultimo autore o "corresponding author". La produzione scientifica nel periodo di osservazione ha coinvolto 26 gruppi di ricerca afferenti a 21 strutture Universitarie o Ospedaliere. Dall'analisi dei dati è emersa una correlazione tra il numero di soci afferenti allo stesso gruppo di ricerca ed il numero di lavori prodotti. Dei 189 lavori selezionati, 156 (83 %) sono stati pubblicati su riviste con IF mentre i restanti 33 (17 %) sono stati pubblicati su riviste censite da PubMed ma senza IF. l'IF complessivo nel periodo di osservazione è stato di 541.528 (media di 3.47 ± 1.81), mediana di 3.093, range 0.899-12.047.

L'andamento temporale della produzione scientifica e dell'IF/anno ha mostrato una tendenziale deflessione del numero dei lavori/anno: 64 nel 2014, 53 nel 2015, 46 nel 2016 e 26 nei primi 5 mesi del 2017. Per verificare l'impatto dei lavori selezionati è stato valutato il numero delle citazioni tramite Scopus. Relativamente alle pubblicazioni 2014 non è stata rilevata alcuna correlazione tra l'IF e numero di citazioni. Le prime 18 riviste scientifiche nelle quali sono stati pubblicati i lavori coprono il 46% della produzione globale. Relativamente alle malattie metaboliche oggetto di pubblicazione, il 43.5 % è costituito da malattie lisosomiali e, nell'ambito di queste, la Malattia di Fabry è risultata la più frequentemente riportata.

Sono state valutate le istituzioni con maggior numero di lavori pubblicati e non sempre è stata riscontrata una correlazione tra il numero dei lavori ed IF complessivo/medio. I gruppi più rappresentativi sono risultati essere gli ospedali pediatrici ed i grandi Dipartimenti di Pediatria. Dai lavori censiti emerge una chiara tendenza alla collaborazione tra i diversi centri Italiani con frequente riscontro di co-authorship da soci provenienti da diverse strutture. Questo punto, che documenta l'esistenza di un network italiano nell'ambito della SIMMESN, rappresenta un elemento portante del contributo scientifico italiano al settore delle malattie metaboliche.

È stata inoltre valutata la partecipazione dei soci SIMMESN alla produzione di linee guida internazionali e/o consensus conference: 8 linee guida internazionali pubblicate su importanti riviste (range di IF 3,08-16,321) aventi come oggetto errori congeniti della rimetilazione, deficit di cistationina beta-sintetasi (omocistinuria classica), galattosemia, fenilchetonuria, glutarico aciduria tipo I, metilmalonico e propionico acidemia, e deficit di sfingomielinasi acida hanno avuto almeno 1 socio/coautore tra gli "opinion leader".

La co-authorship dei soci SIMMESN a pubblicazioni aventi come primo/ultimo autore o "corresponding authors" ricercatori italiani non-soci SIMMESN è risultata rilevante, soprattutto nell'ambito delle malattie lisosomiali, malattie mitocondriali, deficit di Alfa 1- antitripsina, terapia genica, malattie mitocondriali, sviluppo di metodologie per terapie intratecale, trapianti di cellule staminali, metodologie di screening neonatali.

AFFILIAZIONI SOCI

40 Centri Italiani

Centri con ruolo "leader" nelle pubblicazioni 2014-2017

26 gruppi di ricerca afferenti a 21 centri Ospedalieri/Universitari

Pubblicazioni

Division of Metabolism - Bambino Gesù Children's Hospital, Rome , Italy

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Ipertrigliceridemia infantile transitoria	**Dionisi-Vici C,** Shteyer E, Niceta M, Rizzo C, Pode-Shakked B, Chillemi G, Bruselles A, Semeraro M, Barel O, Eyal E, Kol N, Haberman Y, Lahad A, Diomedi-Camassei F, Marek-Yagel D, Rechavi G, Tartaglia M, Anikster Y.	Expanding the molecular diversity and phenotypic spectrum of glycerol 3-phosphate dehydrogenase 1 deficiency	J Inherit Metab Dis. Sep;39(5):689-95.	2016	3.541	
Malattie Perossisomiali	Semeraro M, Rizzo C, Boenzi S, Cappa M, Bertini E, Antonetti G, **Dionisi-Vici C.**	A new multiplex method for the diagnosis of peroxisomal disorders allowing simultaneous determination of plasma very-long-chain fatty acids, phytanic,pristanic,docosahexaenoic and bile acids by high-performance liquid chromatography-atmospheric pressure chemical ionization-tandem mass spectrometry.	Clin Chim Acta. Jul 1;458:159-64	2016	2.799	
Encefalopatia Etilmalonico	**Dionisi-Vici C**, Diodato D, Torre G, Picca S, Pariante R, Giuseppe Picardo S, Di Meo I, Rizzo C, Tiranti V, Zeviani M, De Ville De Goyet J	Liver transplant in ethylmalonic encephalopathy: a new treatment for an otherwise fatal disease.	Brain. Apr;139(Pt4):1045-51.	2016	10.103	
Metabolismo colesterolo	Boenzi S, Deodato F, Taurisano R, Goffredo BM, Rizzo C**, Dionisi-Vici C.**	Evaluation of plasma cholestane-3β,5α,6β-triol and 7-ketocholesterol in inherited disorders related to cholesterol metabolism	J Lipid Res. Mar;57(3):361-7**.**	2016	4.368	

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Tirosinemia	Maiorana A, Malamisura M, Emma F, Boenzi S, Di Ciommo VM**, Dionisi-Vici C.**	Early effect of NTBC on renal tubular dysfunction in hereditary tyrosinemia type 1	Mol Genet Metab. Nov;113(3):188-93.	2014	3.093	
Difetti di CblC	Fischer S, Huemer M, Baumgartner M, Deodato F, Ballhausen D, Boneh A, Burlina AB, Cerone R, Garcia P, Gökçay G, Grünewald S, Häberle J, Jaeken J, Ketteridge D, Lindner M, Mandel H, Martinelli D, Martins EG, Schwab KO, Gruenert SC, Schwahn BC, Sztriha L, Tomaske M, Trefz F, Vilarinho L, Rosenblatt DS, Fowler B, **Dionisi-Vici C.**	Clinical presentation and outcome in a series of 88 patients with the cblC defect.	J Inherit Metab Dis. Sep;37(5):831-40.	2014	3.365	

dihydrolipoamide dehydrogenase deficiency	Carrozzo R, Torraco A, Fiermonte G, Martinelli D, Di Nottia M, Rizza T, Vozza A, Verrigni D, Diodato D, Parisi G, Maiorana A, Rizzo C, Pierri CL, Zucano S, Piemonte F, Bertini E, **Dionisi-Vici C.**	Riboflavin responsive mitochondrial myopathy is a new phenotype of dihydrolipoamide dehydrogenase deficiency. The chaperon-like effect of vitamin B2.	Mitochondrion.Sep;18:49-57.	2014	3.249	
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Malattia mitocondriale	Catteruccia M, Verrigni D, Martinelli D, Torraco A, Agovino T, Bonafé L, D'Amico A, Donati MA, Adorisio R, Santorelli FM, Carrozzo R, Bertini E, **Dionisi-Vici C**	Persistent pulmonary arterial hypertension in the newborn (PPHN): a frequent manifestation of TMEM70 defective patients.	Mol Genet Metab. Mar;111(3):353-9.	2014	3.093	
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Difetti di CblC	Pastore A, Martinelli D, Piemonte F, Tozzi G, Boenzi S, Di Giovamberardino G, Petrillo S, Bertini E, **Dionisi-Vici C.**	Glutathione metabolism in cobalamin deficiency type C (cblC).	J Inherit Metab Dis.Jan;37(1):125-9.	2014	3.365	

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Malattia lisosomiale Deficit lipasi acida	Taurisano R, Maiorana A, De Benedetti F, Dionisi-Vici C, Boldrini R, **Deodato F**.	Wolman disease associated with hemophagocytic lymphohistiocytosis: attempts for an explanation.	Eur J Pediatr. Oct;173(10):1391-4	2014	1.791	
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Ricerca di base	Semeraro M, Muraca M, Catesini G, Inglese R, Iacovone F, Barraco GM, Manco M, Boenzi S, Dionisi-Vici C, **Rizzo C**.	Determination of plasma pipecolic acid by an easy and rapid liquid chromatography-tandem mass spectrometry method.	Clin Chim Acta. Feb 2;440:108-12.	2015	2,799	
Malattia lisosomiale Gangliosidosi GM1	**Deodato F**, Procopio E, Rampazzo A, Taurisano R, Donati MA, Dionisi-Vici C, Caciotti A, Morrone A, Scarpa M.	The treatment of juvenile/adult GM1-gangliosidosis with Miglustat may reverse disease progression.	Metab Brain Dis. Jun 3.	2017	2.063	

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Galattosemia	Viggiano E, Marabotti A, Politano L, **Burlina A.**	Galactose-1-Phosphate Uridyltransferase deficiency: a literature review of the putative mechanisms of short and long-term complications and allelic variants.	Clin Genet. Apr 4.	2017	3.892	

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Difetti neurotrasmettitori	**Burlina AB**, Celato A, Polo G, Edini C, Burlina AP.	The Utility of CSF for the Diagnosis of Primary and Secondary Monoamine Neurotransmitter Deficiencies.	EJIFCC. Mar 8;28(1):64	2017	/	
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NPC	Polo G, Burlina A, Furlan F, Kolamunnage T, Cananzi M, Giordano L, Zaninotto M, Plebani M, **Burlina A**.	High level of oxysterols in neonatal cholestasis: a pitfall in analysis of biochemical markers for Niemann-Pick type C disease.	Clin Chem Lab Med. Jul 1;54(7):1221-9.	2016	3.017	
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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
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Malattia lisosomiale Malattia di Fabry	Politei JM, Bouhassira D, Germain DP, Goizet C, Guerrero-Sola A, Hilz MJ, Hutton EJ, Karaa A, Liguori R, Üçeyler N, Zeltzer LK, **Burlina A.**	Pain in Fabry Disease: Practical Recommendations for Diagnosis and Treatment.	CNS Neurosci Ther.Jul;22(7):568-76.	2016	4.019	
Malattia lisosomiale Malattia di Fabry	Kolodny E, Fellgiebel A, Hilz MJ, Sims K, Caruso P, Phan TG, Politei J, Manara R, **Burlina A.**	Cerebrovascular involvement in Fabry disease: current status of knowledge.	Stroke. 2015 Jan;46(1):302-13.	2015	5.787	
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Malattia mitocondriale Sindrome di Leigh	Minoia F, Bertamino M, Picco P, Severino M, Rossi A, Fiorillo C, Minetti C, Nesti C, Santorelli FM, **Di Rocco M**.	Widening the Heterogeneity of Leigh Syndrome: Clinical, Biochemical, and Neuroradiologic Features in a Patient Harboring a NDUFA10 Mutation.	JIMD Rep. Mar 1.	2017	/	
Malattia lisosomiale Malattia di Gaucher	Madeo A, Garaventa A, Sementa AR, Suffia C, **Di Rocco M**.	The unusual association between Neuroblastoma and Gaucher Disease:ase report and review of the literature.	Blood Cells Mol Dis. Nov 23. pii: S1079-9796(16)30184-X.	2016	2.731	

Malattia lisosomiale Malattia di Pompe	Pichiecchio A, Rossi M, Cinnante C, Colafati GS, De Icco R, Parini R, Menni F, Furlan F, Burlina A, Sacchini M, Donati MA, Fecarotta S, Casa RD, Deodato F, Taurisano R, **Di Rocco M**.	Muscle MRI of classic infantile pompe patients: Fatty substitution and edema-like changes.	Muscle Nerve. Sep 26	2016	2.713.	
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NPC	**Di Rocco M**, Barone R, Madeo A, Fiumara A.	Miglustat Does Not Prevent Neurological Involvement in Niemann Pick C Disease.	Pediatr Neurol. Oct;53(4):e15	2015	1.866	
Fibrodisplasia progressiva	Bertamino M, Severino M, Schiaffino MC, Garrè ML, Bocciardi R, Ravazzolo R, Rossi A, **Di Rocco M**.	New insights into central nervous system involvement in FOP: Case report and review of the literature.	Am J Med Genet A. Nov;167A(11):2817-21.	2015	2.082	
Malattia lisosomiale Malattia di Gaucher	**Di Rocco M**, Andria G, Deodato F, Giona F, Micalizzi C, Pession A.	Early diagnosis of Gaucher disease in pediatric patients: proposal for a diagnostic algorithm.	Pediatr Blood Cancer. Nov;61(11):1905-9.	2014	2.634	
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Metachromatic leukodystrophy	Siri L, Rossi A, Lanza F, Mazzotti R, Costa A, Stroppiano M, Gaiero A, Cohen A, Biancheri R, **Filocamo M.**	A novel homozygous splicing mutation in PSAP gene causes metachromatic leukodystrophy in two Moroccan brothers.	Neurogenetics. 15:101-106	2014	3.426	
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Mucolipidosis IV	Mirabelli-Badenier M, Severino M, Tappino B, Tortora D, Camia F, Zanaboni C, Brera F, Priolo E, Rossi A, Biancheri R, Di Rocco M**, Filocamo M.**	A novel homozygous MCOLN1 double mutant allele leading to TRP channel domain ablation underlies Mucolipidosis IV in an Italian Child.	Metab Brain Dis. 30:681-6.	2015	2.6	
Varie malattie rare	Baldo C, Casareto L, Renieri A, Merla G, Garavaglia B, Goldwurm S, Pegoraro E, Moggio M, Mora M, Politano L, Sangiorgi L, Mazzotti R, Viotti V, Meloni I, Pellico MT, Barzaghi C, Chiuhui Mary Wang CM, Monaco L and **Filocamo M.**	The alliance between genetic biobanks and patient organisations: the experience of the Telethon Network of Genetic Biobanks.	OJRD 11:142.	2016	3.290	
Gaucher disease	Amico G, Grossi S, Vijzelaar R, Lanza F, Mazzotti R, Corsolini F, Ketemab M, **Filocamo M.**	MLPA-Based Approach for Initial and Simultaneous Detection of GBA Deletions and Recombinant Alleles in Patients Affected by Gaucher Disease.	Mol Genet Metab. 119: 329–337	2016	3.093	

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
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Deficit α lipoproteina	Sechi A, Dardis A, Zampieri S, Rabacchi C, Zanoni P, Calandra S, De Maglio G, Pizzolitto S, Maruotti V, Di Muzio A, Platt F, **Bembi B**.	Effects of miglustat treatment in a patient affected by an atypical form of Tangier disease.	Orphanet J Rare Dis. Sep 18;9:143.	2014	3.358	

Malattia Lisosomiale Malattia di Gaucher	Sechi A, Deroma L, Dardis A, Ciana G, Bertin N, Concolino D, Linari S, Perria C, **Bembi B**	Long term effects of enzyme replacement therapy in an Italian cohort of type 3 Gaucher patients.	Mol Genet Metab. Nov;113(3):2138	2014	2.625	
Malattia Lisosomiale Malattia di Fabry	Sechi A, Nucifora G, Piccoli G, Dardis A, **Bembi B**.	Myocardial fibrosis as the first sign of cardiac involvement in a male patient with Fabry disease: report of a clinical case and discussion on the utility of the magnetic resonance in Fabry pathology.	BMC Cardiovasc Disord. Jul 16;14:86.	2014	1.5	
Malattia lisosomiale NPC	**Dardis A**, Zampieri S, Canterini S, Newell KL, Stuani C, Murrell JR, Ghetti B, Fiorenza MT, Bembi B, Buratti E.	Altered localization and functionality of TAR DNA Binding Protein 43 (TDP-43) in niemann- pick disease type C.	Acta Neuropathol Commun. May 18;4(1):52.	2016	/	
Malattia lisosomiale NPC	Romanello M, Zampieri S, Bortolotti N, Deroma L, Sechi A, Fiumara A, Parini R, Borroni B, Brancati F, Bruni A, Russo CV, Bordugo A, Bembi B, **Dardis A**.	Comprehensive Evaluation of Plasma 7-Ketocholesterol and Cholestan-3β,5α,6β-Triol in an Italian Cohort of Patients Affected by Niemann-Pick Disease due to NPC1 and SMPD1 Mutations.	Clin Chim Acta. Apr 1;455:39-45.	2016	2.799	
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Malattia lisosomiale	Malini E, Zampieri S, Deganuto M, Romanello M, Sechi A, Bembi B, **Dardis A**.	Role of LIMP-2 in the intracellular trafficking of β-glucosidase in different human cellular models.	FASEB J. Sep;29(9):3839-52	2015	5.299	
Malattia lisosomiale NPC	Zampieri S, Bianchi E, Cantile C, Saleri R, Bembi B, **Dardis A**.	Characterization of a spontaneous novel mutation in the NPC2 gene in a cat affected by Niemann Pick type C disease.	PLoS One. Nov 14;9(11):e112503.	2014	3.234.	
Malattia lisosomiale	**Dardis A**, Zanin I, Zampieri S, Stuani C, Pianta A, Romanello M, Baralle FE, Bembi B, Buratti E.	Functional characterization of the common c.-32-13T>G mutation of GAA gene: identification of potential therapeutic agents.	Nucleic Acids Res.Jan;42(2):1291-302	2014	9.112	

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
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Malattia lisosomiale Malattia di Gaucher	Giuffrida G, Lombardo R, Di Francesco E, Parrinello L, Di Raimondo F, **Fiumara A**.	Successful switch from enzyme replacement therapy to miglustat in an adult patient with type 1 Gaucher disease: a case report.	J Med Case Rep. Nov 8;10(1):315.	2016	/	
CDGs	**Fiumara A**, Barone R, Del Campo G, Striano P, Jaeken J.	Electroclinical Features of Early-Onset Epileptic Encephalopathies in Congenital Disorders of Glycosylation (CDGs).	JIMD Rep. ;27:93-9	2016	/	
NPC	Di Rocco M, Barone R, Madeo A, **Fiumara A**.	Miglustat Does Not Prevent Neurological Involvement in Niemann Pick C Disease.	Pediatr Neurol. Oct;53(4):e15.	2015	1.866	
CDG	**Barone R**, Carrozzi M, Parini R, Battini R, Martinelli D, Elia M, Spada M, Lilliu F, Ciana G, Burlina A, Leuzzi V, Leoni M, Sturiale L, Matthijs G, Jaeken J, Di Rocco M, Garozzo D, **Fiumara A**.	A nationwide survey of PMM2-CDG in Italy: high frequency of a mild neurological variant associated with the L32R mutation.	J Neurol. Jan;262(1):154-64.	2015	3.408	
	Barone R, Fichera M, De Grandi M, Battaglia M, Lo Faro V, Mattina T, Rizzo R	Familial 18q12.2 deletion supports the role of RNA-binding protein CELF4 in autism spectrum disorders.	Am J Med Genet A. pr 13	2017	2.082	

Gangliosidosi GM2	**Barone R**, Sturiale L, Fiumara A, Palmigiano A, Bua RO, Rizzo R, Zappia M, Garozzo D.	CSF N-glycan profile reveals sialylation deficiency in a patient with GM2 gangliosidosis presenting as childhood disintegrative disorder.	Autism Res. Apr;9(4):423-8.	2016	3.048	
CDG	**Barone R**, Fiumara A, Jaeken J.	Congenital disorders of glycosylation with emphasis on cerebellar involvement.	Semin Neurol. Jul;34(3):357-66.	2014	1.838	

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Malattia lisosomiale Malattia di Fabry	Ferri L, Covello G, Caciotti A, Guerrini R, Denti MA, **Morrone A**..	Double-target Antisense U1snRNAs Correct Mis-splicing Due to c.639+861C>T and c.639+919G>A GLA Deep Intronic Mutations.	Mol Ther Nucleic Acids. Oct 25;5(10):e380	2016	5.048	
Sindrome di Barth	Ferri L, Donati MA, Funghini S, Cavicchi C, Pensato V, Gellera C, Natacci F, Spaccini L, Gasperini S, Vaz FM, Cooper DN, Guerrini R, **Morrone A**.	Intra-individual plasticity of the TAZ gene leading to different heritable mutations in siblings with Barth syndrome.	Eur J Hum Genet. Dec;23(12):1708-12.	2015	4.58	
Malattia lisosomiale MPS IV A	Caciotti A, Tonin R, Rigoldi M, Ferri L, Catarzi S, Cavicchi C, Procopio E, Donati MA, Ficcadenti A, Fiumara A, Barone R, Garavelli L, Rocco MD, Filocamo M, Antuzzi D, Scarpa M, Mooney SD, Li B, Skouma A, Bianca S, Concolino D, Casalone R, Monti E, Pantaleo M, Giglio S, Guerrini R, Parini R, **Morrone A**.	Optimizing the molecular diagnosis of GALNS: novel methods to define and characterize Morquio-A syndrome-associated mutations.	Hum Mutat. Mar;36(3):357-68	2015	5.089	

Malattia lisosomiale MPS IV A	**Morrone A**, Caciotti A, Atwood R, Davidson K, Du C, Francis-Lyon P, Harmatz P, Mealiffe M, Mooney S, Oron TR, Ryles A, Zawadzki KA, Miller N.	Morquio A syndrome-associated mutations: a review of alterations in the GALNS gene and a new locus-specific database.	Hum Mutat. Nov;35(11):1271-9.	2014	5.089	
Difetti ciclo urea OTC	Cavicchi C, Donati M, Parini R, Rigoldi M, Bernardi M, Orfei F, Gentiloni Silveri N, Colasante A, Funghini S, Catarzi S, Pasquini E, la Marca G, Mooney S, Guerrini R, **Morrone A**.	Sudden unexpected fatal encephalopathy in adults with OTC gene mutations-Clues for early diagnosis and timely treatment.	Orphanet J Rare Dis. Jul 16;9:105.	2014	3.290	
Malattia lisosomiale MPS IV A	**Morrone A**, Tylee KL, Al-Sayed M, Brusius-Facchin AC, Caciotti A, Church HJ, Coll MJ, Davidson K, Fietz MJ, Gort L, Hegde M, Kubaski F, Lacerda L, Laranjeira F, Leistner-Segal S, Mooney S, Pajares S, Pollard L, Ribeiro I, Wang RY, Miller N.	Molecular testing of 163 patients with Morquio A (Mucopolysaccharidosis IVA) identifies 39 novel GALNS mutations.	Mol Genet Metab. Jun;112(2):160-70	2014	3.093	
deficienza di Aminoacilasi I	Ferri L, Funghini S, Fioravanti A, Biondi EG, la Marca G, Guerrini R, Donati MA, **Morrone A**.	Aminoacylase I deficiency due to ACY1 mRNA exon skipping.	Clin Genet. Oct;86(4):367-72.	2014	3.931 (14)	
Malattia lisosomiale Malattia di Pompe	Sacchini M, Procopio E, Pasquini E, Pochiero F, Ombrone D, LaMarca G, Catarzi S, Morrone A, **Donati MA.**	Alpha Glucosidase Assay on Dried Blood Spot in the Early Diagnosis of Infantile Pompe Disease.	J Neuromuscul Dis. 2(s1):S53.	2015	/	
Screening neonatale	**la Marca G**, Giocaliere E, Malvagia S, Villanelli F, Funghini S, Ombrone D, Della Bona M, Forni G, Canessa C, Ricci S, Romano F, Guerrini R, Resti M, Azzari C.	Development and validation of a 2nd tier test for identification of purine nucleoside phosphorylase deficiency patients during expanded newborn screening by liquid chromatography-tandem mass spectrometry.	Clin Chem Lab Med. Apr;54(4):627-32.	2016	3.017	
Screening neonatale	Ombrone D, Giocaliere E, Forni G, Malvagia S, **la Marca G**.	Expanded newborn screening by mass spectrometry: New tests, future perspectives.	Mass Spectrom Rev. Jan-Feb;35(1):71-84	2016	9.346	

Screening neonatale	Malvagia S, Haynes CA, Grisotto L, Ombrone D, Funghini S, Moretti E, McGreevy KS, Biggeri A, Guerrini R, Yahyaoui R, Garg U, Seeterlin M, Chace D, De Jesus VR, **la Marca G**.	Heptadecanoylcarnitine (C17) a novel candidate biomarker for newborn screening of propionic and methylmalonic acidemias.	Clin Chim Acta. Oct 23;450:342-8.	2015	2.799	
Screening neonatale	Villanelli F, Giocaliere E, Malvagia S, Rosati A, Forni G, Funghini S, Shokry E, Ombrone D, Della Bona ML, Guerrini R, **la Marca G**.	Dried blood spot assay for the quantification of phenytoin using Liquid Chromatography-Mass Spectrometry.	Clin Chim Acta. Feb 2;440:31-5	2015	2.799	
Screening neonatale	Shokry E, Villanelli F, Malvagia S, Rosati A, Forni G, Funghini S, Ombrone D, Della Bona M, Guerrini R, **la Marca G**	Therapeutic drug monitoring of carbamazepine and its metabolite in children from dried blood spots using liquid chromatography and tandem mass spectrometry.	J Pharm Biomed Anal. May 10;109:164-70	2015	3.169	
Screening neonatale	**la Marca G**.	Newborn Screening: Are We Ready for It?	J Neuromuscul Dis.;2(s1):S10.	2015	/	
Screening neonatale	**la Marca G**, Canessa C, Giocaliere E, Romano F, Malvagia S, Funghini S, Moriondo M, Valleriani C, Lippi F, Ombrone D, Della Bona ML, Speckmann C, Borte S, Brodszki N, Gennery AR, Weinacht K, Celmeli F, Pagel J, de Martino M, Guerrini R, Wittkowski H, Santisteban I, Bali P, Ikinciogullari A, Hershfield M, Notarangelo LD, Resti M, Azzari C.	Diagnosis of immunodeficiency caused by a purine nucleoside phosphorylase defect by using tandem mass spectrometry on dried blood spots.	J Allergy Clin Immunol. Jul;134(1):155-9.	2014	12.047	
Screening neonatale	**la Marca G**, Giocaliere E, Malvagia S, Funghini S, Ombrone D, Della Bona ML, Canessa C, Lippi F, Romano F, Guerrini R, Resti M, Azzari C.	The inclusion of ADA-SCID in expanded newborn screening by tandem mass spectrometry.	J Pharm Biomed Anal. 2014 Jan;88:201-6	2014	2.947	
Screening neonatale	**la Marca G**.	Mass spectrometry in clinical chemistry: the case of newborn screening.	J Pharm Biomed Anal. 2014 Dec;101:174-82	2014	2.947	

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
	Melis D, Carbone F, Minopoli G, La Rocca C, Perna F, De Rosa V, Galgani M, Andria G, Parenti G, Matarese G.	Cutting Edge: Increased Autoimmunity Risk in Glycogen Storage Disease Type 1b Is Associated with a Reduced Engagement of Glycolysis in T Cells and an Impaired Regulatory T Cell Function.	J Immunol. 2017 May 15;198(10):3803-3808.	2017	4.9	
	Cappuccio G, Vitiello F, Casertano A, Fontana P, Genesio R, Bruzzese D, Ginocchio VM, Mormile A, Nitsch L, Andria G, **Melis D**.	New insights in the interpretation of array-CGH: autism spectrum disorder and positive family history for intellectual disability predict the detection of pathogenic variants.	Ital J Pediatr. 2016 Apr 12;42:39	2016	1.61	
	Melis D, Rossi A, Pivonello R, Del Puente A, Pivonello C, Cangemi G, Negri M, Colao A, Andria G, **Parenti G.**	Reduced bone mineral density in glycogen storage disease type III: evidence for a possible connection between metabolic imbalance and bone homeostasis.	Bone. 2016 May;86:79-85.	2016	3.7	
	Melis D, Rossi A, Pivonello R, Salerno M, Balivo F, Spadarella S, Muscogiuri G, Della Casa R, Formisano P, Andria G, Colao A, **Parenti G.**	Glycogen storage disease type Ia (GSDIa) but not Glycogen storage disease type Ib (GSDIb) is associated to an increased risk of metabolic syndrome: possible role of microsomal glucose 6-phosphate accumulation.	Orphanet J Rare Dis. 2015 Jul 29;10:91	2015	3.29	
	Melis D,Minopoli G, Balivo F, Marcolongo P, Parini R, Paci S, Dionisi-Vici C, Della Casa R, Benedetti A, Andria G, **Parenti G.**	Vitamin E Improves Clinical Outcome of Patients Affected by Glycogen Storage Disease Type Ib.	JIMD Rep. 2016;25:39-45.	2016	/	

	Fecarotta S, Romano A, Della Casa R, Del Giudice E, Bruschini D, Mansi G, Bembi B, Dardis A, Fiumara A, Di Rocco M, Uziel G, Ardissone A, Roccatello D, Alpa M, Bertini E, D'Amico A, Dionisi-Vici C, Deodato F, Caviglia S, Federico A, Palmeri S, Gabrielli O, Santoro L, Filla A, Russo C, Parenti G, **Andria G.**	Long term follow-up to evaluate the efficacy of miglustat treatment in Italian patients with Niemann-Pick disease type C.	Orphanet J Rare Dis. 2015 Feb 27;10:22.	2015	3,29	
	Parenti G, Andria G, Valenzano KJ.	Pharmacological Chaperone Therapy: Preclinical Development, Clinical Translation, and Prospects for the Treatment of Lysosomal Storage Disorders.	MolTher. 2015 Jul;23(7):1138-48.	2015	6.93	
	Scala I,Concolino D, Della Casa R, Nastasi A, Ungaro C, Paladino S, Capaldo B, Ruoppolo M, Daniele A, Bonapace G, Strisciuglio P, Parenti G, **Andria G.**	Long-term follow-up of patients with phenylketonuria treated with tetrahydrobiopterin: a seven years experience.	Orphanet J Rare Dis. 2015 Feb 8;10:14	2015	3.29	
	Melis D,Cozzolino M, Minopoli G, Balivo F, Parini R, Rigoldi M, Paci S, Dionisi-Vici C, Burlina A, Andria G, **Parenti G.**	Progression of renal damage in glycogen storage disease type I is associated to hyperlipidemia: a multicenter prospective Italian study.	J Pediatr. 2015 Apr;166(4):1079-82	2015	3.89	
	Parenti G, Andria G, Ballabio A.	Lysosomal storage diseases: from pathophysiology to therapy.	Annu Rev Med. 2015;66:471-86.	2015	10.95	
	Parenti G, Moracci M, Fecarotta S, **Andria G.**	Pharmacological chaperone therapy for lysosomal storage diseases	Future Med Chem. 2014 Jun;6(9):1031-45	2014	3.74	

	Parenti G, Fecarotta S, la Marca G, Rossi B, Ascione S, Donati MA, Morandi LO, Ravaglia S, Pichiecchio A, Ombrone D, Sacchini M, Pasanisi MB, De Filippi P, Danesino C, Della Casa R, Romano A, Mollica C, Rosa M, Agovino T, Nusco E, Porto C, **Andria G**.	A chaperone enhances blood α-glucosidase activity in Pompe disease patients treated with enzyme replacement therapy.	MolTher. 2014 Nov;22(11):2004-12.	2014	6.22	
	Melis D, Della Casa R, Balivo F, Minopoli G, Rossi A, Salerno M, Andria G, **Parenti G.**	Involvement of endocrine system in a patient affected by glycogen storage disease 1b: speculation on the role of autoimmunity.	Ital J Pediatr. 2014 Mar 19;40(1):30.	2014	1.52	
	Melis D,Pivonello R, Cozzolino M, Della Casa R, Balivo F, Del Puente A, Dionisi-Vici C, Cotugno G, Zuppaldi C, Rigoldi M, Parini R, Colao A, Andria G, **Parenti G.**	Impaired bone metabolism in glycogen storage disease type 1 is associated with poor metabolic control in type 1a and with granulocyte colony-stimulating factor therapy in type 1b.	Horm Res Paediatr. 2014;81(1):55-62.	2014	1.57	
	Strisciuglio P, Concolino D.	New Strategies for the Treatment of Phenylketonuria (PKU).	Metabolites. 2014 Nov 4;4(4):1007-17.	2014	/	
	Concolino D, Mascaro I, Moricca MT, Bonapace G, Matalon K, Trapasso J, Radhakrishnan G, Ferrara C, Matalon R, Strisciuglio P.	Long-term treatment of phenylketonuria with a new medical food containing large neutral amino acids.	Eur J Clin Nutr. 2017 Jan;71(1):51-55.	2017	2.935	

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Malattie lisosomiali MPS	**Gabrielli O**, Zampini L, Monachesi C, Marchesiello RL, Padella L, Santoro L, Volpi N, Concolino D, Fiumara A, Rigon L, Mazzoli M, Carnielli VP, Giovagnoni A, Catassi C, Galeazzi T, Coppa GV.	Early diagnosis of mucopolysaccharidoses in developing countries: A low cost and easy execution approach.	Clin Chim Acta. Feb 28;468:150-151.	2017	2.799	
Malattie lisosomiali MPS	**Gabrielli O**, Clarke LA, Ficcadenti A, Santoro L, Zampini L, Volpi N, Coppa GV.	12 year follow up of enzyme-replacement therapy in two siblings with attenuated mucopolysaccharidosis I: the important role of early treatment.	BMC Med Genet. 2016 Mar 10;17:19.	2016	2.094	
	Coppa GV, Facinelli B, Magi G, Marini E, Zampini L, Mantovani V, Galeazzi T, Padella L, Marchesiello RL, Santoro L, Coscia A, Peila C, Volpi N, **Gabrielli O**.	Human milk glycosaminoglycans inhibit in vitro the adhesion of Escherichia coli and Salmonella fyris to human intestinal cells.	Pediatr Res. 2016 Apr;79(4):603-7.	2006	2.761	
Malattie lisosomiali Deficit slfatasi	Volpi N, Coppa GV, Zampini L, Maccari F, Galeotti F, Garavelli L, Galeazzi T, Padella L, Santoro L, **Gabrielli O**.	Plasmatic and urinary glycosaminoglycan profile in a patient affected by multiple sulfatase deficiency.	Clin Chem Lab Med. 2015 Jun;53(7):e157-60.	2015	3.017	
Malattie lisosomiali Deficit solfatasi	Garavelli L, Santoro L, Iori A, Gargano G, Braibanti S, Pedori S, Melli N, Frattini D, Zampini L, Galeazzi T, Padella L, Pepe S, Wischmeijer A, Rosato S, Ivanovski I, Iughetti L, Gelmini C, Bernasconi S, Superti-Furga A, Ballabio A, **Gabrielli O.**	Multiple sulfatase deficiency with neonatal manifestation.	Ital J Pediatr. Dec 17;40:86	2014	1.614	

Malattie lisosomiali MPS	**Zampini L**, Padella L, Marchesiello RL, Santoro L, Monachesi C, Giovagnoni A, Catassi C, **Gabrielli O**, Coppa GV, Galeazzi T.	Importance of the combined urinary procedure for the diagnosis of Mucopolysaccharidoses.	Clin Chim Acta. 2017 Jan;464:165-169.	2017	2.799	

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Malattie lisosomiali	Tasegian A, Paciotti S, Ceccarini MR, Codini M, Moors T, Chiasserini D, Albi E, Winchester B, van de Berg WDJ, Parnetti L, **Beccari T**.	Origin of α-mannosidase activity in CSF.	Int J Biochem Cell Biol. Mar 27;87:34-37.	2017	4.046	Base
Malattie lisosomiali	Paciotti S, Codini M, Tasegian A, Ceccarini MR, Cataldi S, Arcuri C, Fioretti B, Albi E, **Beccari T**.	Lysosomal alpha-mannosidase and alpha-mannosidosis.	Front Biosci (Landmark Ed). 2017 Jan 1;22:157-167.	2017	2.5	Base
Malattie lisosomiali	Chiasserini D, Paciotti S, Eusebi P, Persichetti E, Tasegian A, Kurzawa-Akanbi M, Chinnery PF, Morris CM, Calabresi P, Parnetti L, **Beccari T**.	Selective loss of glucocerebrosidase activity in sporadic Parkinson's disease and dementia with Lewy bodies.	Mol Neurodegener. Mar 27;10:15.	2015	6.510	
Malattie lisosomiali	Persichetti E, Chiasserini D, Parnetti L, Eusebi P, Paciotti S, De Carlo C, Codini M, Tambasco N, Rossi A, El-Agnaf OM, Calabresi P, **Beccari T**.	Factors influencing the measurement of lysosomal enzymes activity in human cerebrospinal fluid.	PLoS One. Jul 1;9(7):e101453	2014	3.234	
Malattie lisosomiali	Paciotti S, Persichetti E, Klein K, Tasegian A, Duvet S, Hartmann D, Gieselmann V, **Beccari T**.	Accumulation of free oligosaccharides and tissue damage in cytosolic α-mannosidase (Man2c1)-deficient mice.	J Biol Chem. Apr 4;289(14):9611-22	2014	4.258	

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Tisoninemia	**Leuzzi V**, Mastrangelo M, Giannini MT, Carbonetti R, Hoffmann GF.	Neuromotor and cognitive outcomes of early treatment in tyrosine hydroxylase deficiency type B.	Neurology. 2017 Jan 31;88(5):501-502.	**2017**	8.166 (15)	
	Mastrangelo M, Scheffer IE, Bramswig NC, Nair LD, Myers CT, Dentici ML, Korenke GC, Schoch K, Campeau PM, White SM, Shashi V, Kansagra S, Van Essen AJ, **Leuzzi V**.	Epilepsy in KCNH1-related syndromes.	Epileptic Disord. 2016 Jun 1;18(2):123-36	**2016**	0.942	
	Leuzzi V, Rossi L, Gabucci C, Nardecchia F, Magnani M.	Erythrocyte-mediated delivery of recombinant enzymes.	J Inherit Metab Dis. 2016 Jul;39(4):519-30	**2016**	3.541	
PKU	Manti F, Nardecchia F, Chiarotti F, Carducci C, Carducci C, **Leuzzi V**.	Psychiatric disorders in adolescent and young adult patients with phenylketonuria.	Mol Genet Metab. 2016 Jan;117(1):12-8.	**2016**	3,093	
PKU	Mastrangelo M, Chiarotti F, Berillo L, Caputi C, Carducci C, Di Biasi C, Manti F, Nardecchia F, **Leuzzi V**.	The outcome of white matter abnormalities in early treated phenylketonuric patients: A retrospective longitudinal long-term study.	Mol Genet Metab. 2015 Nov;116(3):171-7	**2015**	3,093	
PKU	Carducci C, Santagata S, Friedman J, Pasquini E, Carducci C, Tolve M, Angeloni A, **Leuzzi V**.	Urine sepiapterin excretion as a new diagnostic marker for sepiapterin reductase deficiency.	Mol Genet Metab. 2015 Aug;115(4):157-60.	**2015**	3,093	
PKU	Nardecchia F, Manti F, Chiarotti F, Carducci C, Carducci C, **Leuzzi V**.	Neurocognitive and neuroimaging outcome of early treated young adult PKU patients: A longitudinal study.	Mol Genet Metab. 2015 Jun-Jul;115(2-3):84-90.	**2015**	3,093	

	Leuzzi V, Micheli R, D'Agnano D, Molinaro A, Venturi T, Plebani A, Soresina A, Marini M, Ferremi Leali P, Quinti I, Pietrogrande MC, Finocchi A, Fazzi E, Chessa L, Magnani M.	Positive effect of erythrocyte-delivered dexamethasone in ataxia-telangiectasia.	Neurol Neuroimmunol Neuroinflamm. 2015 Apr 9;2(3):e98	**2015**	/	
PKU	**Leuzzi V**, Mannarelli D, Manti F, Pauletti C, Locuratolo N, Carducci C, Carducci C, Vanacore N, Fattapposta F.	Age-related psychophysiological vulnerability to phenylalanine in phenylketonuria.	Front Pediatr. 2014 Jun 23;2:57	**2014**	/	
	Leuzzi V, Mastrangelo M, Polizzi A, Artiola C, van Kuilenburg AB, Carducci C, Ruggieri M, Barone R, Tavazzi B, Abeling NG, Zoetekouw L, Sofia V, Zappia M, Carducci C.	Report of two never treated adult sisters with aromatic L-amino Acid decarboxylase deficiency: a portrait of the natural history of the disease or an expanding phenotype?	JIMD Rep. 2015;15:39-45.	**2015**	/	
	Mastrangelo M, Bernasconi P, De Liso P, Caputi C, Bertino S, **Leuzzi V**.	Teaching video neuroimages: clinical course of infantile ascending hereditary spastic paralysis.	Neurology. 2014 Feb 18;82(7):e61.	**2014**	8.166	
NLC	**Leuzzi V**, Garavaglia B, Manti F, Bertino S, Nardocci N.	Running apraxia as a presenting symptom of neuronal ceroid lipofuscinosis 6.	Mov Disord. 2014 Feb;29(2):277-8.	**2014**	6.01	
Deficit BH4	Nardecchia F, Chiarotti F, Carducci C, Santagata S, Valentini G, Angeloni A, Blau N, **Leuzzi V**.	Altered tetrahydrobiopterin metabolism in patients with phenylalanine hydroxylase deficiency.	Eur J Pediatr. 2017 May 24.	**2017**	1.921	
Deficit BH4	Santagata S, Di Carlo E, Carducci Ca, Angeloni A, Leuzzi V, **Carducci C**	Development of a new UPLC-ESI-MS/MS method for the determination of biopterin and neopterin in dried blood spot	Clin Chim Acta 2017 Mar;466:145-151.	**2017**	2,799	

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
MPSI	**Parini R**, Jones SA, Harmatz PR, Giugliani R, Mendelsohn NJ.	The natural history of growth in patients with Hunter syndrome: Data from the Hunter Outcome Survey (HOS).	Mol Genet Metab. 2016 Apr;117(4):438-46.	2016	3.09	
Fabry	**Parini R**, Schiffmann R, Fotheringham I, Todorova L.	A Systematic Review of The Humanistic Burden of Disease In Patients With Fabry Disease.	Value Health. 2015 Nov;18(7):A762	2015	3.824	
Glicogenosi III	Brambilla A, Mannarino S, Pretese R, Gasperini S, Galimberti C, **Parini R**.	Improvement of Cardiomyopathy After High-Fat Diet in Two Siblings with Glycogen Storage Disease Type III.	JIMD Rep. 2014;17:91-5.	2014	/	
Ciclo urea	Grioni D, Furlan F, Canonico F, **Parini R**.	Epilepsia partialis continua and generalized nonconvulsive status epilepticus during the course of argininemia: a report on two cases.	Neuropediatrics. 2014 Apr;45(2):123-8	2014	1.29	
Fabry	Rigoldi M, Concolino D, Morrone A, Pieruzzi F, Ravaglia R, Furlan F, Santus F, Strisciuglio P, Torti G, **Parini R**.	Intrafamilial phenotypic variability in four families with Anderson-Fabry disease.	Clin Genet. 2014 Sep;86(3):258-63.	2014	3.931	
Fabry	Concolino D, Degennaro E, **Parini R**;	Delphi consensus on the current clinical and therapeutic knowledge on Anderson-Fabry disease.	Eur J Intern Med. 2014 Oct;25(8):751-6	2014	2.591	
MPS	**Parini R**, Deodato F, Di Rocco M, Lanino E, Locatelli F, Messina C, Rovelli A, Scarpa M.	Open issues in Mucopolysaccharidosis type I-Hurler.	Orphanet J Rare Dis. 2017 Jun 15;12(1):112.	2017	3.507	

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
PKU	Moretti F[1], Pellegrini N[2], Salvatici E[3], Rovelli V[3], Banderali G[3], Radaelli G[3], Scazzina F[2], **Giovannini M**[4], Verduci E[5].	Dietary glycemic index, glycemic load and metabolic profile in children with phenylketonuria	NutrMetabCardiovasc Dis. 2017 Feb;27(2):176-182.	**2017**	**2015 3.390**	
PKU	Verduci E**[1]**, Banderali G[2], Moretti F[3], Lassandro C[4], Cefalo G[5], Radaelli G[6], Salvatici E[7], **Giovannini M[8].**	Diet in children with phenylketonuria and risk of cardiovascular disease: A narrative overview	NutrMetabCardiovasc Dis. 2016 Mar;26(3):171-7	**2016**	**3.390**	
GSD I - III	Allegrini D[1], Autelitano A[2], Fogagnolo P[2], De Cillà S[3], Piozzi E[4], Mazza M[4], **Paci S**[5], Montanari C[5], Riva E[5], Rossetti L[2].	Lens opacities in glycogenoses type I and III	Can J Ophthalmol. 2015 Dec;50(6):480-4.	**2015**	**1.460**	
PKU	**Giovannini M[1]**, Riva E, Salvatici E, Cefalo G, Radaelli G.	Randomized controlled trial of a protein substitute with prolonged release on the protein status of children with phenylketonuria	J Am CollNutr. 2014;33(2):103-10. d	**2014**	**2.245**	

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
LSD	**Scarpa M**, Bellettato CM, Lampe C, Begley DJ.	Neuronopathic lysosomal storage disorders: Approaches to treat the central nervous system.	Best Pract Res Clin Endocrinol Metab. 2015 Mar;29(2):159-71	2015	5.070	
LSD	Mazzoccoli G, Mazza T, Vinciguerra M, Castellana S, **Scarpa M**.	The biological clock and the molecular basis of lysosomal storage diseases.	JIMD Rep. 2015;18:93-105	2015	/	

MPSII	Zanetti A, Tomanin R, Rampazzo A, Rigon C, Gasparotto N, Cassina M, Clementi M, **Scarpa M**.	A Hunter Patient with a Severe Phenotype Reveals Two Large Deletions and Two Duplications Extending 1.2 Mb Distally to IDS Locus.	JIMD Rep. 2014	2014	/	
MPSII	Manara R, Concolino D, Rampazzo A, Zanetti A, Tomanin R, Faggin R, **Scarpa M**.	Chiari 1 malformation and holocord syringomyelia in hunter syndrome.	JIMD Rep. 2014;12:31-5	2014	/	
MPS II	Tomanin R, Zanetti A, D'Avanzo F, Rampazzo A, Gasparotto N, Parini R, Pascarella A, Concolino D, Procopio E, Fiumara A, Borgo A, Frigo AC, Scarpa M.	Clinical efficacy of enzyme replacement therapy in paediatric Hunter patients, an independent study of 3.5 years.	Orphanet J Rare Dis. 2014 Sep 18;9:129	2014	3.358	

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1 **Nome MR**	2 **Autori**	3 **Titolo del lavoro**	4 **Rivista, volume, pagine**	5 **Anno**	6 **I. F.**	7 **Note**
Fabry	**Spada M**, Kasper D, Pagliardini V, Biamino E, Giachero S, Porta F.	Metabolic progression to clinical phenotype in classic Fabry disease.	Ital J Pediatr. 2017 Jan 3;43(1):1	**2017**	**1.614**	
Fabry	Mignani R, Pieruzzi F, Berri F, Burlina A, Chinea B, Gallieni M, Pieroni M, Salviati A, **Spada M**.	FAbry STabilization indEX (FASTEX): an innovative tool for the assessment of clinical stabilization in Fabry disease.	Clin Kidney J. 2016 Oct;9(5):739-47.	**2016**	**/**	
PKU	Porta F, Ponzone A, **Spada M**.	Long-term safety and effectiveness of pramipexole in tetrahydrobiopterin deficiency.	Eur J Paediatr Neurol. 2016 Nov;20(6):839-842.	**2016**	**1.923**	
PKU	Porta F, Ponzone A, **Spada M**.	Target Prolactin Range in Treatment of Tetrahydrobiopterin Deficiency.	J Pediatr. 2016 Jan;168:236-9.e1	**2016**	**3.890**	
MMA	**Spada M**, Calvo PL, Brunati A, Peruzzi L, Dell'Olio D, Romagnoli R, Porta F.	Liver transplantation in severe methylmalonic acidemia: The sooner, the better.	J Pediatr. 2015 Nov;167(5):1173.	**2015**	**3.890**	

MMA	**Spada M**, Calvo PL, Brunati A, Peruzzi L, Dell'Olio D, Romagnoli R, Porta F.	Early Liver Transplantation for Neonatal-Onset Methylmalonic Acidemia.	Pediatrics. 2015 Jul;136(1):e252-6.	**2015**	**5.473**	
Galattosemia	Porta F, Pagliardini S, Pagliardini V, Ponzone A, **Spada M**.	Newborn screening for galactosemia: a 30-year single center experience.	World J Pediatr. May;11(2):160-4	**2015**	**1.025**	
PKU Bh4	Porta F, Ponzone A, **Spada M**.	Short prolactin profile for monitoring treatment in BH4 deficiency.	Eur J Paediatr Neurol. 2015 May;19(3):360-3.	**2015**	**1.923**	
Pompe	Porta F, Pagliardini V, Roasio L, Biamino E, **Spada M**.	Playing competitive basketball in face of late-onset pompe disease.	Muscle Nerve. Feb;51(2):302-3.	**2015**	**2.713**	
OTC	D'Onofrio V, Poma F, Enea A, Santarelli F, Lovera C, **Spada M**.	Hyperammonemic coma in a patient with late-onset OTC deficiency.	Pediatr Med Chir. 2014 Jun 30;36(3):9.	**2014**	**/**	
	Porta F, Mussa A, Baldassarre G, Perduca V, Farina D, Spada M, Ponzone .A	Genealogy of breastfeeding.	**E** ur J Pediatr. 2016 Jan;175(1):105-12	**2016**	**1.791**	
	Mussa A, Baldassarre G, **Porta F**.	Fracture odds and body mass index in children	J Pediatr. 2014 Dec;165(6):1274.	**2014**	**3.890**	
Pompe	**Mongini T**, Vercelli L, Ponzalino V, Bortolani S.	Early Recognition of Late-Onset Pompe Disease by Skeletal Muscle Signs and Symptoms.	J Neuromuscul Dis. 2015;2(s1):S2. No abstract available	**2015**	**/**	
	Mongini T, Toscano A.	Foreword.	J Neuromuscul Dis. 2015;2(s1):Si-Sii	**2015**	**/**	

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Organico-aciduria	Villani GR, Gallo G, Scolamiero E, Salvatore F, **Ruoppolo M**.	"Classical organic acidurias": diagnosis and pathogenesis.	Clin Exp Med. 2016 Sep 9.	**2016**	**2.854**	

MMA	Caterino M, Chandler RJ, Sloan JL, Dorko K, Cusmano-Ozog K, Ingenito L, Strom SC, Imperlini E, Scolamiero E, Venditti CP, **Ruoppolo M**.	The proteome of methylmalonic acidemia (MMA): the elucidation of altered pathways in patient livers.	Mol Biosyst. 2016 Feb;12(2):566-74.	**2016**	**2.829**	
	Ruoppolo M, Scolamiero E, Caterino M, Mirisola V, Franconi F, Campesi I.	Female and male human babies have distinct blood metabolomic patterns.	Mol Biosyst. 2015 Sep;11(9):2483-92.	**2015**	**2.829**	
Screening neonatale	Scolamiero E, Cozzolino C, Albano L, Ansalone A, Caterino M, Corbo G, di Girolamo MG, Di Stefano C, Durante A, Franzese G, Franzese I, Gallo G, Giliberti P, Ingenito L, Ippolito G, Malamisura B, Mazzeo P, Norma A, Ombrone D, Parenti G, Pellecchia S, Pecce R, Pierucci I, Romanelli R, Rossi A, Siano M, Stoduto T, Villani GR, Andria G, Salvatore F, Frisso G, **Ruoppolo M**.	Targeted metabolomics in the expanded newborn screening for inborn errors of metabolism.	Mol Biosyst. 2015 Jun;11(6):1525-35.	**2015**	**2.829**	
cblC	Caterino M, Pastore A, Strozziero MG, Di Giovamberardino G, Imperlini E, Scolamiero E, Ingenito L, Boenzi S, Ceravolo F, Martinelli D, Dionisi-Vici C, **Ruoppolo M**.	The proteome of cblC defect: in vivo elucidation of altered cellular pathways in humans.	J Inherit Metab Dis. 2015 Sep;38(5):969-79.		**3.541**	
	Ruoppolo M, Campesi I, Scolamiero E, Pecce R, Caterino M, Cherchi S, Mercuro G, Tonolo G, Franconi F.	Serum metabolomic profiles suggest influence of sex and oral contraceptive use.	Am J Transl Res. 2014 Oct 11;6(5):614-24.	**2014**	**3.402**	
Screening neonatale	Scolamiero E, Villani GR, Ingenito L, Pecce R, Albano L, Caterino M, di Girolamo MG, Di Stefano C, Franzese I, Gallo G, **Ruoppolo M**.	Maternal vitamin B12 deficiency detected in expanded newborn screening.	Clin Biochem. 2014 Dec;47(18):312-7.	**2014**	**2.275**	
	Caterino M, Aspesi A, Pavesi E, Imperlini E, Pagnozzi D, Ingenito L, Santoro C, Dianzani I, **Ruoppolo M**.	Analysis of the interactome of ribosomal protein S19 mutants.	Proteomics. 2014 Oct;14(20):2286-96.	**2014**	**4,079**	

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
cblA	Maines E, Morandi G, Gugelmo G, Ion-Popa F, Campostrini N, Pasini A, Vincenzi M, Teofoli F, Camilot M, **Bordugo A**.	Vitamin B12 Administration by Subcutaneous Catheter Device in a Cobalamin A (cblA) Patient.	JIMD Rep. 2016	2016	/	
Altro	Maines E, Morandi G, Olivieri F, Camilot M, Cavarzere P, Gaudino R, Antoniazzi F, **Bordugo A**.	Erratum to: Growth Hormone Deficiency and Lysinuric Protein Intolerance: Case Report and Review of the Literature.	JIMD Rep. 2015	2015	/	
Altro	**Maines E**, Baggio L, Gugelmo L, Morandi G, Bordugo A	Newborn with rhizomelia and difficult breathing	Skeletal radiology Feb 46(2):291-292	2017	1.19	
Altro	**Tadiotto E,** Maines E, Degani D, banzato C, balter R, Gugelmo G, dardis A, Giordano G, Bordugo A	A Neonate with abdominal distention and failure to thrive	Arch Dis Educ Pract Ed 26 Apr		1.33	
Altro	**Bordugo A**; Carlin E, Demarini S, Faletra F., Colonna F.	A neonate with a milky blood: what can it be?	Arch Dis Child Fetal Neonatal Ed 99(6) F514	2014	3,96	

Unità operativa / Dipartimento /Azienda: Clinical Psychology, Department of Health Sciences, University of Milan, Milan, Italy

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
PKU	**Borghi L**, Salvatici E, Riva E, Giovannini M, Vegni EA.	Psychological and psychosocial implications for parenting a child with phenylketonuria: a systematic review.	Minerva Pediatr. 2017 May 4.	2017	/	

altro	Vegni E, **Borghi L**.	Patient-centered medicine in palliative cancer care: the impossible choice between physician's responsibility and patient's preferences.	Tumori. 2017 Mar 24;103(2):e21.	2017	/	
altro	**Borghi L**, Tesoro V, Vegni E, Bini T, Leone D.	A qualitative content analysis of HIV consultation: when conversation is about aother than disease].	Recenti Prog Med. 2016 Mar;107(3):149-56.	**2016**	**/**	
altro	Meschi T, Ticinesi A, Prati B, Montali A, Ventura A, Nouvenne A, **Borghi L**.	A novel organizational model to face the challenge of multimorbid elderly patients in an internal medicine setting: a case study from Parma Hospital, Italy.	Intern Emerg Med. 2016 Aug;11(5):667-76.	2016	2.340	
altro	**Borghi L**, Johnson I, Barlascini L, Moja EA, Vegni E.	Do occupational therapists' communication behaviours change with experience?	Scand J Occup Ther. 2016;23(1):50-6.	2016	0.957	
Omocisteinemia	Lippi G, Mattiuzzi C, Meschi T, Cervellin G, **Borghi L**.	Homocysteine and migraine. A narrative review.	Clin Chim Acta. 2014 Jun 10;433:5-11.	2014	2.824	
altro	Lippi G, **Borghi L**.	A short story on how the H-index may change the fate of scientists and scientific publishing.	Clin Chem Lab Med 2014 Feb;52(2):e1-3.	2014	3.017	

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1 **Nome MR**	2 **Autori**	3 **Titolo del lavoro**	4 **Rivista, volume, pagine**	5 **Anno**	6 **I. F.**	7 **Note**
Malattia di Gaucher	Ceravolo F, Grisolia M, Sestito S, Falvo F, Moricca MT, **Concolino D**.	Combination therapy in a patient with chronic neuronopathic Gaucher disease: a case report.	J Med Case Rep. 2017 Jan 20;11(1):19.	**2017**	**/**	
Malattia di Gaucher	Sestito S, Filocamo M, Ceravolo F, Falvo F, Grisolia M, Moricca MT, Cantaffa R, Grossi S, Strisciuglio P, **Concolino D**.	Norrbottnian clinical variant of Gaucher disease in Southern Italy.	J Hum Genet. 2017 Apr;62(4):507-511	**2017**	**2.487**	

PKU	**Concolino D**, Mascaro I, Moricca MT, Bonapace G, Matalon K, Trapasso J, Radhakrishnan G, Ferrara C, Matalon R, Strisciuglio P.	Long-term treatment of phenylketonuria with a new medical food containing large neutral amino acids.	Eur J Clin Nutr. 2017 Jan;71(1):51-55.	**2017**	**2.935**	
Malattia di Fabry	Pensabene L, Sestito S, Nicoletti A, Graziano F, Strisciuglio P, **Concolino D**.	Gastrointestinal Symptoms of Patients with Fabry Disease.	Gastroenterol Res Pract. 2016;2016:9712831.	2016	/	
Malattia di Fabry	Di Martino MT, Scionti F, Sestito S, Nicoletti A, Arbitrio M, Hiram Guzzi P, Talarico V, Altomare F, Sanseviero MT, Agapito G, Pisani A, Riccio E, Borrelli O, **Concolino D°**, Pensabene L°.	Genetic variants associated with gastrointestinal symptoms in Fabry disease.	Oncotarget. 2016 Dec 27;7(52):85895-85904	2015	5.008	
PKU	Strisciuglio P, **Concolino D**.	New Strategies for the Treatment of Phenylketonuria (PKU).	Metabolites. 2014 Nov 4;4(4):1007-17.	2014	/	
Malattia di Fabry	**Concolino D**, Degennaro E, Parini R; Fabry Delphi working group.; Fabry Delphi working group..	Delphi consensus on the current clinical and therapeutic knowledge on Anderson-Fabry disease.	Eur J Intern Med. 2014 Oct;25(8):751-6.	2014	2.591	
	Ceravolo F, Messina S, Rodolico C, Strisciuglio P, **Concolino D**.	Myoglobinuria as first clinical sign of a primary alpha-sarcoglycanopathy.	Eur J Pediatr. 2014 Feb;173(2):239-42.	2014	1.791	

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
	Muccilli V, Cardullo N, Spatafora C, Cunsolo V, Tringali C.	α-Glucosidase inhibition and antioxidant activity of an oenological commercial tannin. Extraction, fractionation and analysis by HPLC/ESI-MS/MS and (1)H NMR.	Food Chem. 2017 Jan 15;215:50-60.	**2017**	**4.052**	

	Muccilli V, Saletti R, Cunsolo V, Ho J, Gili E, Conte E, Sichili S, Vancheri C, Foti S.	Protein profile of exhaled breath condensate determined by high resolution mass spectrometry.	J Pharm Biomed Anal. 2015 Feb;105:134-49	**2015**	**3.169**	

Department of Movement, Human and Health Sciences, Unit of Biology, Genetics and Biochemistry, Università degli Studi di Roma "Foro Italico", Rome, Italy

1 **Nome MR**	**2** **Autori**	**3** **Titolo del lavoro**	**4** **Rivista, volume, pagine**	**5** **Anno**	**6** **I. F.**	**7** **Note**
	Duranti G, Ceci R, Sgrò P, Sabatini S, Di Luigi L.	Influence of the PDE5 inhibitor tadalafil on redox status and antioxidant defense system in C2C12 skeletal muscle cells.	Cell Stress Chaperones. 2017 May;22(3):389-396.	**2017**	**2.583**	

AO Pediatria-Pession- Dipartimento di Scienze Mediche e Chirurgiche - AOU Sant'Orsola-Malpighi ,- Bologna

1 **Nome MR**	**2** **Autori**	**3** **Titolo del lavoro**	**4** **Rivista, volume, pagine**	**5** **Anno**	**6** **I. F.**	**7** **Note**
Glicogenosi tipo II (M.Pompe)	Ortolano R, Baronio F, Masetti R, Prete A, **Cassio A,** Pession A.	Letter to the Editors: Concerning "Divergent clinical outcomes of alphaglucosidase enzyme replacement therapy in two siblings with infantile-onset Pompe disease treated in the symptomatic or pre-symptomatic state" by Takashi M et al.	Mol Genet Metab Rep. 2017 Mar 7;11:1.	**2017**	**/**	

Trial clinici farmacologici

Stato dell'arte sullo sviluppo dei farmaci orfani

Numerosi ricercatori SIMMESN hanno partecipato a *trial* clinici con farmaci orfani nell'ambito delle malattie metaboliche ereditarie.
I dati sono stati raccolti attraverso la compilazione di una scheda inviata a tutti i soci SIMMENS.
Dei 19 Centri clinici italiani specializzati in malattie metaboliche ereditarie e afferenti alla SIMMENS (http://www.sismme.it/centriclinici/italia.html), 15 (70%) hanno compilato il questionario e comunicato i dati. Globalmente, il censimento 2014-2017 ha consentito di individuare 40 partecipazioni a trial clinici (media di 3.63±4.34 studi per centro). Le malattie lisosomiali hanno rappresentato il target principale di trial con farmaci orfani (70% degli studi), seguite dalle aminoacidopatie (27.5%) e dalle malattie mitocondriali (2.5%). Dal punto di vista del meccanismo d'azione terapeutica, i farmaci più utilizzati sono risultati gli enzimi ricombinanti (55%), seguiti da molecole con attività di attivazione o inibizione enzimatica (inclusa la riduzione del substrato) (35%) e da altri approcci (10%: cellule staminali, terapia genica, riduzione dello stress ossidativo e altro).

Trial clinici farmacologici

1 Nome MR	2 *Sponsor*	3 *Partners*		4 Ruolo		5 Denominazione del *trial*	6 Farmaco	7 Durata		8 Note
		I	N	C	P			C	T (anno)	
Niemann-Pick B	Sanofi						Terapia enzimatica sostitutiva			S.O.C. Centro di Coordinamento Regionale per le Malattie Rare, Azienda Ospedaliero Universitaria Santa Maria della Misericordia, Udine
Niemann-Pick B	Sanofi						Terapia enzimatica sostitutiva			S.O.C. Centro di Coordinamento Regionale per le Malattie Rare, Azienda Ospedaliero Universitaria Santa Maria della Misericordia, Udine
Niemann-Pick C	Orphazyme						Chaperone			S.O.C. Centro di Coordinamento Regionale per le Malattie Rare, Azienda Ospedaliero Universitaria Santa Maria della Misericordia, Udine

Pompe	Genzyme						Terapia enzimatica sostitutiva			S.C Pediatria I - Presidio "Ospedale Infantile Regina Margherita, Azienda Ospedaliero-Universitaria Città della Salute e della Scienza di Torino
Pompe	Indipendente						Terapia enzimatica sostitutiva			S.C Pediatria I - Presidio "Ospedale Infantile Regina Margherita, Azienda Ospedaliero-Universitaria Città della Salute e della Scienza di Torino
Tirosinemia	Sobi						Inibizione enzimatica			S.C Pediatria I - Presidio "Ospedale Infantile Regina Margherita, Azienda Ospedaliero-Universitaria Città della Salute e della Scienza di Torino
Iperammoniemie neonatali	Indipendente						Cellule staminali epatiche			S.C Pediatria I - Presidio "Ospedale Infantile Regina Margherita, Azienda Ospedaliero-Universitaria Città della Salute e della Scienza di Torino

Alcaptonuria	Sobi						Inibizione enzimatica			S.C Pediatria I - Presidio "Ospedale Infantile Regina Margherita, Azienda Ospedaliero-Universitaria Città della Salute e della Scienza di Torino
Fabry	Genzyme						Terapia enzimatica sostitutiva			S.C Pediatria I - Presidio "Ospedale Infantile Regina Margherita, Azienda Ospedaliero-Universitaria Città della Salute e della Scienza di Torino
Gaucher	Genzyme						Inibizione substrato			S.C Pediatria I - Presidio "Ospedale Infantile Regina Margherita, Azienda Ospedaliero-Universitaria Città della Salute e della Scienza di Torino
Tirosinemia	Sobi						Inibizione enzimatica			Centro HUB rete Malattie Metaboliche Ereditarie Area Vasta Emilia Nord - Emilia Romagna - Piacenza
Niemann-Pick C	Orphazyme						Chaperone			Unità Operativa Complessa di Patologia Metabolica, Ospedale Pediatrico Bambino Gesù IRCCS, Roma

Sindome di Leigh	EDISON Farmaceutical						Riduzione stress ossidativo			Unità Operativa Complessa di Patologia Metabolica, Ospedale Pediatrico Bambino Gesù IRCCS, Roma
Niemann-Pick C	Aifa						Riduzione substrato			Dipartimento di Scienze mediche Traslazionali- Sezione di Pediatria-Università "Federico II"- Napoli (Coordinatore)
Pompe	Telethon UILDM						Terapia combinata ERT+Chaperon			Dipartimento di Scienze mediche Traslazionali- Sezione di Pediatria-Università "Federico II"- Napoli
MPS VI	Telethon UE						Terapia genica			Dipartimento di Scienze mediche Traslazionali- Sezione di Pediatria-Università "Federico II"- Napoli
Wolman	SynagevaAlexion						Terapia enzimatica sostitutiva			Dipartimento di Scienze mediche Traslazionali- Sezione di Pediatria-Università "Federico II"- Napoli
PKU	Aifa						Chaperone			Dipartimento di Scienze mediche Traslazionali- Sezione di Pediatria-Università "Federico II"- Napoli (Coordinatore)

MPS IVA	Biomarin						Terapia enzimatica sostitutiva			Unità Operativa Semplice Malattie Metaboliche Rare, Clinica Pediatrica, Fondazione MBBM, Azienda Ospedaliera San Gerardo, Monza
MPS IVA	Biomarin						Terapia enzimatica sostitutiva			Unità Operativa Semplice Malattie Metaboliche Rare, Clinica Pediatrica, Fondazione MBBM, Azienda Ospedaliera San Gerardo, Monza
MPS IVA	Biomarin						Terapia enzimatica sostitutiva			Unità Operativa Semplice Malattie Metaboliche Rare, Clinica Pediatrica, Fondazione MBBM, Azienda Ospedaliera San Gerardo, Monza
MPS IIIA	Shire						Terapia enzimatica sostitutiva			Unità Operativa Semplice Malattie Metaboliche Rare, Clinica Pediatrica, Fondazione MBBM, Azienda Ospedaliera San Gerardo, Monza
MPS IIIA	Shire						Terapia enzimatica sostitutiva			Unità Operativa Semplice Malattie Metaboliche Rare, Clinica Pediatrica, Fondazione MBBM, Azienda Ospedaliera San Gerardo, Monza

MPS II	Shire						Terapia enzimatica sostitutiva			Unità Operativa Semplice Malattie Metaboliche Rare, Clinica Pediatrica, Fondazione MBBM, Azienda Ospedaliera San Gerardo, Monza
Fabry	Shire						Terapia enzimatica sostitutiva			Unità Operativa Semplice Malattie Metaboliche Rare, Clinica Pediatrica, Fondazione MBBM, Azienda Ospedaliera San Gerardo, Monza
MPS I	Genzyme						Terapia enzimatica sostitutiva			Unità Operativa Semplice Malattie Metaboliche Rare, Clinica Pediatrica, Fondazione MBBM, Azienda Ospedaliera San Gerardo, Monza
Pompe	Genzyme						Terapia enzimatica sostitutiva			Unità Operativa Semplice Malattie Metaboliche Rare, Clinica Pediatrica, Fondazione MBBM, Azienda Ospedaliera San Gerardo, Monza
MPS VI	Biomarin						Terapia enzimatica sostitutiva			Unità Operativa Semplice Malattie Metaboliche Rare, Clinica Pediatrica, Fondazione MBBM, Azienda Ospedaliera San Gerardo, Monza

Tirosinemia	Sobi						Inibizione enzimatica			Unità Operativa Semplice Malattie Metaboliche Rare, Clinica Pediatrica, Fondazione MBBM, Azienda Ospedaliera San Gerardo, Monza
MPS IVA	Biomarin						Terapia enzimatica sostitutiva			Unità Operativa Semplice Malattie Metaboliche Rare, Clinica Pediatrica, Fondazione MBBM, Azienda Ospedaliera San Gerardo, Monza
Niemann-Pick C	Orphazyme									Unità Operativa Semplice Malattie Metaboliche Rare, Clinica Pediatrica, Fondazione MBBM, Azienda Ospedaliera San Gerardo, Monza
MPS IIIB	Alexion									Unità Operativa Semplice Malattie Metaboliche Rare, Clinica Pediatrica, Fondazione MBBM, Azienda Ospedaliera San Gerardo, Monza
Pompe	Erasmus MC						Terapia enzimatica sostitutiva			Unità Operativa Semplice Malattie Metaboliche Rare, Clinica Pediatrica, Fondazione MBBM, Azienda Ospedaliera San Gerardo, Monza

PKU	Biomarin						Chaperon			Centro Regionale di riferimento per gli screening neonatali e la diagnosi delle malattie metaboliche, Istituto Giannina Gaslini, Genova
MPS IVA	Biomarin						Terapia enzimatica sostitutiva			DIPARTIMENTO DI PEDIATRIAUNIVERSITA' di CATANIA - Centro di Riferimento Regionale per la cura ed il controllo delle Malattie Metaboliche ereditarie dell'infanzia
Pompe	Indipendente						Terapia enzimatica sostitutiva			DIPARTIMENTO DI PEDIATRIAUNIVERSITA' di CATANIA - Centro di Riferimento Regionale per la cura ed il controllo delle Malattie Metaboliche ereditarie dell'infanzia
PKU	Biomarin						Chaperone			Centro Clinico Regionale di riferimento per diagnosi e follow-up Malattie Metaboliche, Dipartimento di Scienze Mediche e Chirurgiche, A.O.U. S'ORSOLA MALPIGHI BOLOGNA

PKU	MerkSerono						Chaperone			Centro Clinico Regionale di riferimento per diagnosi e follow-up Malattie Metaboliche, Dipartimento di Scienze Mediche e Chirurgiche, A.O.U. S'ORSOLA MALPIGHI BOLOGNA
PKU	MerkSerono						Chaperone			Centro Clinico Regionale di riferimento per diagnosi e follow-up Malattie Metaboliche, Dipartimento di Scienze Mediche e Chirurgiche, A.O.U. S'ORSOLA MALPIGHI BOLOGNA
MPS II	Shire						Terapia enzimatica sostitutiva			Centro Clinico Regionale di riferimento per diagnosi e follow-up Malattie Metaboliche, Dipartimento di Scienze Mediche e Chirurgiche, A.O.U. S'ORSOLA MALPIGHI BOLOGNA

Partecipazione Nazionale a ERN

Rete MetabERN

Il *network* europeo, partner della Society for the Study of Inborn Error of Metabolism (SSIEM), coinvolge 69 Health Care Providers, certificati e approvati dai Ministeri della Sanità di 18 diversi Paesi dell'Unione Europea. In Italia sono stati identificati 11 centri:

- ASST Monza San Gerardo Hospital (S. Gasparine - A. Piperno),
- Azienda Ospedaliera Universitaria "Federico II", Napoli (G. Parenti - P. Strisciuglio),
- Azienda Ospedaliera Universitaria Integrata di Verona (A. Salviati - A. Bordugo),
- Azienda Ospedaliera Universitaria Senese (A. Federico - M.T. Dotti),
- Azienda Ospedaliero-Universitaria Pisana (F. Santini - G. Ceccarini),
- Azienda Sanitaria Universitaria Integrata di Udine (B. Bembi - G.A. Dardis),

- IRCCS Istituto Giannina Gaslini, Genova (M. di Rocco - R. Cerone),
- Ospedale Pediatrico Meyer, Firenze (M.A. Donati - E. Pasquini),
- IRCCS Ospedale Pediatrico Bambino Gesù, Roma (C. Dionisi-Vici - E. Bertini),
- Ospedale San Paolo, ASST Santi Paolo e Carlo, Milano (G. Cefalo - G. Banderali),
- Università-Ospedale di Padova (A. Burlina - G. Polo).

I *subnetwork* "Lysosomal Diseases", "Carbohydrate, fatty acid Oxidation and ketone Bodies Disorders" e "Disorder of pyruvate metabolism, Krebs cycle defects, mitochondrial oxidative phosphorylation disorders and thiamine transport and metabolism disorders" vedono coinvolti con ruolo di cocordinatore medici provenienti dalla rete dei centri italiani.

Dai dati ufficiali MetabERN il Prof G. Parenti é coordinatore del "Lysosomal Diseases Subnetwork", Carlo Dionisi Vici del subnetwork "Carbohydrate, fatty acid Oxidation and ketone Bodies Disorders", Enrico Bertini del "Disorder of pyruvate metabolism, Krebs cycle defects, mitochondrial oxidative phosphorylation disorders and thiamine transport and metabolism disorders" subnetwork.

HCP	LEADER	VICE	AREA
ASST Monza San Gerardo Hospital	Serena Gasperini	Alberto Piperno	Pediatria Medicina Interna
Azienda Ospedaliera Universitaria "Federico II", Napoli	Giancarlo Parenti	Pietro Strisciuglio	Pediatria
Azienda Ospedaliera Universitaria Integrata di Verona	Alessandro Salviati	Andrea Bordugo	Pediatria
Azienda Ospedaliera Universitaria Senese	Antonio Federico	Maria Teresa Dotti	Neurologia e malattie neuromuscolari
Azienda Ospedaliero-Universitaria Pisana	Ferruccio Santini	Giovanni Ceccarini	Endocrinologia
Azienda Sanitaria Universitaria Integrata di Udine	Bruno Bembi	Andrea Dardis	Centro di Coordinamento Regionale Malattie Rare
Giannina Gaslini Institute (IRCCS Giannina Gaslini)	Maja di Rocco	Roberto Cerone	Ospedale pediatrico
Meyer Children's Hospital	Maria Alice Donati	Elisabetta Pasquini	Ospedale pediatrico
Ospedale Pediatrico Bambino Gesù	Carlo Dionisi Vici	Enrico Bertini	Ospedale Pediatrico
San Paolo Hospital, ASST Santi Paolo e Carlo	Graziella Cefalo	Giuseppe Banderali	Pediatria
University Hospital of Padova	Alberto Burlina	Giulia Polo	Pediatria

Malattie gastroenterologiche e epatologiche pediatriche

A cura della Società Italiana di Gastroenterologia Epatologia e Nutrizione Pediatrica - SIGENP
(Presidente: *Carlo Agostoni*)

Carlo Agostoni
Mara Cananzi
Gabriella Nebbia
Giusy Ranucci

Pubblicazioni

Epatopatie rare pediatriche: pubblicazioni dei gruppi italiani con ruolo *leader*

Per quanto concerne l'attività pubblicativa in ambito epatologico pediatrico, i dati inseriti nel database sono stati ottenuti attraverso una ricerca sulla piattaforma PubMed e selezionando le pubblicazioni inerenti a malattie epatiche rare dell'età pediatrica il cui primo/ultimo autore risulti afferente ad un centro italiano. La ricerca è stata chiusa al 27 giugno 2017. Sono state incluse le pubblicazioni prodotte dal 2014.

Gli inserimenti nel database sono stati distinti per centro di appartenenza del primo/ultimo autore e malattia.

Per quanto concerne la qualità delle pubblicazioni, l'impact factor totale è di circa 252 punti, con un impact factor mediano di 3,95 punti (range 0,5-13,42). La maggior parte della produzione italiana in ambito epatologico pediatrico si concentra sui trapianti di fegato, ma peculiare è la produzione italiana a livello europeo in malattie rare, come la Malattia di Wilson. Negli ultimi 10 anni l'epatologia pediatrica si è espansa in aree cliniche vicine, come le malattie epatiche metaboliche e le condizioni sistemiche con coinvolgimento epatico.

Le malattie epatiche rare generalmnte esordiscono in età pediatrica con quadri di epatite fulminante o possono progredire in maniera subdola fino alla cirrosi epatica. In molti casi quando la malattia è evoluta, l'unico efficace trattamento è il trapianto di fegato. Nei pazienti pediatrici, il ritardo nella diagnosi è responsabile anche di scarsa crescita ed alterazione importante della qualità della vita dei piccoli pazienti.

Molte condizioni epatiche pediatriche sono precursori di malattie epatiche croniche dell'adulto. A tal proposito una sfida importante nell'ambito delle malattie rare del fegato è la transizione delle cure dai pediatri ai medici dell'adulto, considerato che nella maggior parte dei casi i pazienti necessitano di una terapia quotidiana e cronica e hanno una malattia che impatta a livello sistemico sull'organismo, il che rende ancor più complesso il processo di transizione delle cure. In particolare per la gestione clinica e terapeutica della malattia di Wilson e dei disturbi biliari cronici, come l'atresia biliare, che rimane la più comune malattia cronica del fegato pediatrico e l'indicazione per il trapianto di fegato, la medicina dell'adulto ha mutuato molto dell'esperienza fatta in ambito pediatrico. Per questi disordini la ricerca pediatrica ha un ruolo centrale e trainante.

La SIGENP di recente si è fatta promotrice della creazione di un Network Italiano dedicato allo studio dell'Atresia delle vie biliari, in associazione con i neonatolgi ed i chirurghi pediatrici. In particolare è stata promossa la creazione di un registro pilota della malattia. Oltre al fine epidemiologico, il registro ha l'obiettivo di coagulare l'energie dei vari ricercatori coinvolti nello studio della malattia per facilitare le ricerche sulla stessa e anche la gestione di trial clinici multicentrici.

Per quanto concerne la Malattia di Wilson, questa è diventata in ambito epatologico nazionale un modello di malattia rara con ricerche in corso di tipo traslazionale che hanno unito ricercatori di base, clinici e l'associazione dei malati. Le ricerche degli ultimi anni si concentrano sulla genesi di nuove terapie per la cura della malattia.

DI seguito un elenco in via di definizione sulla produzione scientifica degli ultimi anni ricercatori italiani nell'ambito delle malattie rare del fegato in età pediatrica.

Pubblicazioni Italiane sulle epatopatie pediatriche rare

RICERCA CLINICA | 2014-2017 (ultimo triennio)

PRIMO/ULTIMO AUTORE RICERCATORE ITALIANO (ruolo leader)

EPATOPATIE RARE (davvero rare < 1:10.000)

BEST SELLER

Circa 70 pubblicazioni

IF tot: 252

IF mediano di 3,95 punti (range 0,5-13,42)

1. Trapianto di fegato in m.rare
2. Malattia di Wilson

Pubblicazioni

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Gaucher Disease type 2	Gotti G, Marseglia A, De Giacomo C, Iascone M, Sonzogni A, D'Antiga L.	Neonatal Jaundice with Splenomegaly: Not a Common Pick.	Fetal Pediatr Pathol, 35, 108-111	2016	0,5	AO Bergamo
Hepatoblastoma	Saettini F, Conter V, Provenzi M, Rota M, Giraldi E, Foglia C, Cavalleri L, D'Antiga L.	Is multifocality a prognostic factor in childhood hepatoblastoma?	Pediatr Blood Cancer, 61, 1593-7	2014	2,51	
Autoimmune liver disease	Di Giorgio A, Bravi M, Bonanomi E, Alessio G, Sonzogni A, Zen Y, Colledan M, D'Antiga L.	Fulminant hepatic failure of autoimmune aetiology in children.	J Pediatr Gastroenterol Nutr. 2015 Feb;60(2):159-64.	2015	2,4	
Liver transplant	Nacoti M, Corbella D, Fazzi F, Rapido F, Bonanomi E.	Coagulopathy and transfusion therapy in pediatric liver transplantation.	World J Gastroenterol. 2016 Feb 14;22(6):2005-23.	2016	3,8	
Alagille Syndrome	Pavanello M, Severino M, D'Antiga L, Castellan L, Calvi A, Colledan M, Gandolfo C.	Pretransplant management of basilar artery aneurysm and moyamoya disease in a child with Alagille syndrome.	Liver Transpl. 2015 Sep;21(9):1227-30.	2015	3,91	
Non-cirrhotic portal hypertension	D'Antiga L, Dacchille P, Boniver C, Poledri S, Schiff S, Zancan L, Amodio P.	Clues for minimal hepatic encephalopathy in children with noncirrhotic portal hypertension.	J Pediatr Gastroenterol Nutr. 2014 Dec;59(6):689-94.	2014	2,4	

Liver transplant	Marseglia A, Ginammi M, Bosisio M, Stroppa P, Colledan M, D'Antiga L.	Determinants of large drain losses early after pediatric liver transplantation.	Pediatr Transplant. 2017 Apr 17.	2017	1,5	
Liver transplant	Nicastro E, Giovannozzi S, Stroppa P, Casotti V, Callegaro AP, Tebaldi A, Farina C, Colledan M, D'Antiga L.	Effectiveness of Preemptive Therapy for Cytomegalovirus Disease in Pediatric Liver Transplantation.	Transplantation. 2017 Apr;101(4):804-810.	2017	3,67	
Liver carcinogenesis	Ravà M, D'Andrea A, Doni M, Kress TR, Ostuni R, Bianchi V, Morelli MJ, Collino A, Ghisletti S, Nicoli P, Recordati C, Iascone M, Sonzogni A, D'Antiga L, Shukla R, Faulkner GJ, Natoli G, Campaner S, Amati B.	Mutual epithelium-macrophage dependency in liver carcinogenesis mediated by ST18.	Hepatology. 2017 May;65(5):1708-1719.	2017	13,24	
Progressive intrahepatic cholestasis	Iannelli F, Collino A, Sinha S, Radaelli E, Nicoli P, D'Antiga L, Sonzogni A, Faivre J, Buendia MA, Sturm E, Thompson RJ, Knisely AS, Natoli G, Ghisletti S, Ciccarelli FD.	Massive gene amplification drives paediatric hepatocellular carcinoma caused by bile salt export pump deficiency.	Nat Commun. 2015 Jul 2;6:7456.	2015	11,32	
Liver transplant	D'Antiga L, Colledan M.	Surgical gene therapy by domino auxiliary liver transplantation.	Liver Transpl. 2015 Nov;21(11):1338-9.	2015	3,91	

Liver transplant	Colledan M, D'Antiga L.	Biliary complications after pediatric liver transplantation: the endless heel.	Pediatr Transplant. 2014 Dec;18(8):786-7.	2014	1,5	
Bacterial Peritonitis	Di Giorgio A, D'Antiga L.	Diagnosing Spontaneous Bacterial Peritonitis in Children: Tap-in for Higher Scores.	J Pediatr Gastroenterol Nutr. 2017 Feb;64(2):171-172.	2017	2,4	
Neonatal Hemochromatosis	Indolfi G, Bèrczes R, Pelliccioli I, Bosisio M, Agostinis C, Resti M, Zambelli M, Lucianetti A, Colledan M, D'Antiga L.	Neonatal haemochromatosis with reversible pituitary involvement.	Transpl Int, 27, e76-79	2014	3,079	Primo autore Meyer Firenza
Biliary Atresia	Dilena R, Nebbia G, Fiorica L, Farallo M, Degrassi I, Gozzo F, Pelliccia V, Barbieri S, Cossu M, Tassi L	Epilepsy surgery in a liver-transplanted girl with temporal lobe epilepsy and hippocampal sclerosis following PRES with status epilepticus	Eur J Paediatr Neurol, 20(4):652-656	2016	1,91	Fondazione IRCCS Ospedale Maggiore Policlinico Milano
Alagille Syndrome	De Filippis T, Marelli F, Nebbia G, Porazzi P, Corbetta S, Fugazzola L, Gastaldi R, Vigone MC, Biffanti R, Frizziero D, Mandarà L, Prontera P, Salerno M, Maghnie M, Tiso N, Radetti G, Weber G, Persani L	JAG1 Loss-Of-function variations as a novel predisposing event in the pathogenesis of congenital thyroid defects	J Clin Endocrinol Metab, 101(3):861-870	2016	5,51	

Liver damage	Donati B, Motta BM, Pingitore P, Meroni M, Pietrelli A, Alisi A, Petta S, Xing C, Dongiovanni P, del Menico B, Rametta R, Mancina RM, Badiali S, Fracanzani AL, Craxì A, Fargion S, Nobili V, Romeo S, Valenti L.	The rs2294918 E434K variant modulates patatin-like phospholipase domain-containing 3 expression and liver damage.	Hepatology. 2016 Mar;63(3):787-98.	2016	13,24	
Liver transplant	Spada M, Calvo PL, Brunati A, Peruzzi L, Dell'Olio D, Romagnoli R, Porta F.	Early Liver Transplantation for Neonatal-Onset Methylmalonic Acidemia.	Pediatrics. 2015 Jul;136(1):e252-6.	2015	5,2	Ospedale Regina Margherita Torino
Liver transplant	Spada M, Calvo PL, Brunati A, Peruzzi L, Dell'Olio D, Romagnoli R, Porta F.	Liver transplantation in severe methylmalonic acidemia: The sooner, the better.	J Pediatr. 2015 Nov;167(5):1173.	2015	6,1	
Liver transplant	Calvo PL, Serpe L, Brunati A, Nonnato A, Bongioanni D, Olio DD, Pinon M, Ferretti C, Tandoi F, Carbonaro G, Salizzoni M, Amoroso A, Romagnoli R, Canaparo R.	Donor CYP3A5 genotype influences tacrolimus disposition on the first day after paediatric liver transplantation.	Br J Clin Pharmacol. 2017 Jun;83(6):1252-1262.	2017	3,83	

Liver transplant	Bergallo M, Gambarino S, Pinon M, Barat V, Montanari P, Daprà V, Galliano I, Calvo PL.	EBV-encoded microRNAs profile evaluation in pediatric liver transplant recipients.	J Clin Virol. 2017 Jun;91:36-41.	2017		
Congenital Disorder of Glycosilation	Calvo PL, Spada M, Rabbone I, Pinon M, Porta F, Cisarò F, Reggiani S, Cefalù AB, Sturiale L, Garozzo D, Lefeber DJ, Jaeken J.	An Unexplained Congenital Disorder of Glycosylation-II in a Child with Neurohepatic Involvement, Hypercholesterolemia and Hypoceruloplasminemia.	JIMD Rep. 2017 Jun 23.	2017	5,6	
Liver transplant	Calvo PL, Brunati A, Spada M, Romagnoli R, Corso G, Parenti G, Rossi M, Baldi M, Carbonaro G, David E, Pucci A, Amoroso A, Salizzoni M.	Liver transplantation in defects of cholesterol biosynthesis: the case of lathosterolosis.	Am J Transplant. 2014 Apr;14(4):960-5	2014	5,6	
Liver transplant	Calvo PL, Brunati A, Spada M, Romagnoli R, Corso G, Parenti G, Rossi M, Baldi M, Carbonaro G, David E, Pucci A, Amoroso A, Salizzoni M.	Liver transplantation in defects of cholesterol biosynthesis: the case of lathosterolosis.	Am J Transplant. 2014 Apr;14(4):960-5.	2014	5,6	
Liver disease general	Gasparetto M, Pillon M, Cananzi M, Opinto V, Messina C, Guariso G.	Inside and outside the liver: the challenge of hepatitis-associated aplastic anemia in children.	Minerva Pediatr. 2016 Feb;68(1):78-9.	2016		AO Padova
Extra-hepatic Portosystemic Shunt	Rossi G, di Chio T, Nastasio S, Maggiore G.	Spontaneous Extra-hepatic Portosystemic Shunt in Congenital Hepatic Fibrosis.	J Pediatr Gastroenterol Nutr.	2014	2,4	AO Pisa

Giant cell hepatitis with autoimmune hemolytic anemia	Marsalli G, Nastasio S, Sciveres M, Calvo PL, Ramenghi U, Gatti S, Albano V, Lega S, Ventura A, Maggiore G.	Efficacy of intravenous immunoglobulin therapy in giant cell hepatitis with autoimmune hemolytic anemia: A multicenter study.	Clin Res Hepatol Gastroenterol., 40, 83-89	2016	1,98	
Hepatic finrinogen storage disease	Asselta R, Robusto M, Braidotti P, Peyvandi F, Nastasio S, D'Antiga L, Perisic VN, Maggiore G, Caccia S, Duga S	Hepatic fibrinogen storage disease: identification of two novel mutations (p.Asp316Asn, fibrinogen Pisa and p.Gly366Ser, fibrinogen Beograd) impacting on the fibrinogen γ-module.	J Thromb Haemost., 13, 1459-67	2015	5,2	
Giant cell hepatitis with autoimmune hemolytic anemia	Marsalli G, Nastasio S, Sciveres M, Calvo PL, Ramenghi U, Gatti S, Albano V, Lega S, Ventura A, Maggiore G.	Efficacy of intravenous immunoglobulin therapy in giant cell hepatitis with autoimmune hemolytic anemia: A multicenter study.	Clin Res Hepatol Gastroenterol., 40, 83-89	2016	1,98	
Autoimmune hepatitis	Maggiore G, Socie G, Sciveres M, Roque-Afonso AM, Nastasio S, Johanet C, Gottrand F, Fournier-Favre S, Jacquemin E, Bernard O.	Seronegative autoimmune hepatitis in children: Spectrum of disorders.	Dig Liver Dis. 2016 Jul;48(7):785-91.	2016	4,3	
Hepatic finrinogen storage disease	Asselta R, Robusto M, Braidotti P, Peyvandi F, Nastasio S, D'Antiga L, Perisic VN, Maggiore G, Caccia S, Duga S	Hepatic fibrinogen storage disease: identification of two novel mutations (p.Asp316Asn, fibrinogen Pisa and p.Gly366Ser, fibrinogen Beograd) impacting on the fibrinogen γ-module.	J Thromb Haemost., 13, 1459-67	2015	5,2	

congenital intrahepatic Portosystemic Shunt	Paolantonio G, Pietrobattista A, Candusso M, Monti L, de Ville de Goyet J, Torre G, Rollo M.	Congenital double intrahepatic portosystemic shunt: Imaging findings and endovascular closure.	Ann Hepatol, 56, 370-372	2015	2,3	Bambino Gesù Roma
Liver transplant	Dionisi-Vici C, Diodato D, Torre G, Picca S, Pariante R, Giuseppe Picardo S, Di Meo I, Rizzo C, Tiranti V, Zeviani M, De Ville De Goyet J.	Liver transplant in ethylmalonic encephalopathy: a new treatment for an otherwise fatal disease.	Brain, 139, 1045-51	2016	10,2	
Familial Intrahepatic Cholestasis	Giovannoni I, Callea F, Bellacchio E, Torre G, De Ville De Goyet J, Francalanci P.	Genetics and Molecular Modeling of New Mutations of Familial Intrahepatic Cholestasis in a Single Italian Center.	Plos One 2015,:e014	2015	3,2	
congenital intrahepatic Portosystemic Shunt	Bertocchini A, Falappa P, Grimaldi C, Bolla G, Monti L, de Ville de Goyet J.	Intrahepatic portal venous systems in children with noncirrhotic prehepatic portal hypertension: anatomy and clinical relevance.	J Pediatr Surg, 49, 1268-75	2014	1,97	
Hepatic artery aneurysm	Paolantonio G, Tarissi de Jacobis I, Marchesi A, Natali GL, de Ville de Goyet J, Rollo M, Villani A	Right hepatic artery aneurysms in a child with Kawasaki disease: flow-preserving endovascular treatment.	Ann Hepatol, 13, 284-287	2014	2,3	
idiopathic prehepatic portal hypertension	Superina RA, de Ville de Goyet J.	Preemptive Meso-Rex bypass for children with idiopathic prehepatic portal hypertension: trick or treat?	J Pediatr Gastroenterol Nutr., 58, e41	2014	2,4	

Liver transplant	Angelico R, Passariello A, Pilato M, Cozzolino T, Piazza M, Miraglia R, D'Angelo P, Capasso M, Saffioti MC, Alberti D, Spada M.	Ante situm liver resection with inferior vena cava replacement under hypothermic cardiopolmunary bypass for hepatoblastoma: Report of a case and review of the literature.	Int J Surg Case Rep. 2017 Jun 13;37:90-96	2017	2,4	
Liver transplant	De Luca M, Green M, Symmonds J, Klieger SB, Soltys K, Fisher BT.	Invasive candidiasis in liver transplant patients: Incidence and risk factors in a pediatric cohort.	Pediatr Transplant. 2016 Mar;20(2):235-40.	2016	1,5	
Liver transplant	De Ville de Goyet J, di Francesco F, Sottani V, Grimaldi C, Tozzi AE, Monti L, Muiesan P	Splitting livers: Trans-hilar or trans-umbilical division? Technical aspects and comparative outcomes	Pediatr Transplant. 19(5):517-26.	2015	1,44 1	
Liver diseases, general	Hadžić N, Baumann U, McKiernan P, McLin V, Nobili V.	Long-term challenges and perspectives of pre-adolescent liver disease.	Lancet Gastroenterol Hepatol. 2017 Jun;2(6):435-445	2017	10,2	
Liver diseases, general	Della Corte C, Mosca A, Vania A, Alterio A, Alisi A, Nobili V	Pediatric liver diseases: current challenges and future perspectives	Expert Rev Gastroenterol Hepatol. 10(2):255-65.	2016	2,41 7	
Liver cancer	Nobili V, Alisi A, Grimaldi C, Liccardo D, Francalanci P, Monti L, Castellano A, De Ville De Goyet J	Non-alcoholic fatty liver disease and hepatocellular carcinoma in a 7-year-old obese boy: Coincidence or comorbidity?	Pediatr Obes. 9(5):e99-e102.	2014	2,41 9	
Alpha-1-antitrypsin deficiency	Giovannoni I, Callea F, Stefanelli M, Mariani R, Santorelli FM, Francalanci P	Alpha-1-antitrypsin deficiency: from genoma to liver disease. PiZ mouse as model for the development of liver pathology in human	Liver Int. 35(1):198-206.	2015	4,44 7	see METABOLIC

Anomaly of Portal System	Grimaldi C, Adorisio O, di Francesco F, Goyet JV	Congenital Meso-Rex Bypass: A Rare and Remarkable Anatomical Variation of the Portal System	J Pediatr Gastroenterol Nutr. 64(2):e49.	2017	2,625	
Autoimmune liver disease	Cardile S, Alterio T, Candusso M, Pietrobattista A, Liccardo D, Basso MS, Papadatou B, Bracci F, Knafelz D, Torre G.	Autoimmune liver diseases and inflammatory bowel diseases in children: current issues and future perspectives.	Scand J Gastroenterol. 2017 Jun - Jul;52(6-7):662-667.	2017	2,3	
Autoimmune hepatitis	Cardile S, Alterio T, Candusso M, Pietrobattista A, Liccardo D, Basso MS, Papadatou B, Bracci F, Knafelz D, Torre G.	Autoimmune hepatitis and homeopathic therapies: "old wives' tale"	Minerva Pediatr. 66(6):588-90.	2014	0,723	
Autoimmune Hepatitis	Villalta D, Girolami E, Alessio MG, Sorrentino MC, Tampoia M, Brusca I, Daves M, Porcelli B, Barberio G, Conte M, Pantarotto L, Bizzaro N; Study Group on Autoimmune Diseases of the Italian Society of Laboratory Medicine, Italy	Autoantibody Profiling in a Cohort of Pediatric and Adult Patients With Autoimmune Hepatitis	J Clin Lab Anal. 30(1):41-6.	2016	1,549	

Congenital hepatic fibrosis	Locatelli L, Cadamuro M, Spirlì C, Fiorotto R, Lecchi S, Morell CM, Popov Y, Scirpo R, De Matteis M, Amenduni M, Pietrobattista A, Torre G, Schuppan D, Fabris L, Strazzabosco M	Macrophage recruitment by fibrocystin-defective biliary epithelial cells promotes portal fibrosis in congenital hepatic fibrosis	Hepatology. 63(3):965-82.	2016	11,055	First and corresponding authors from University of Milan
Sarcoma of the liver	Merli L, Musini C, Gabor F, Pariente D, Guerin F	Pitfalls in the surgical management of undifferentiated sarcoma of the liver and benefits of preoperative chemotherapy	Eur J Pediatr Surg. 25(1):132-7.	2015	0,98	see PAEDIATRIC CANCERS
Primary biliary cirrhosis	Villalta D, Sorrentino MC, GIROLAMI ELIA, Porcelli B, Barberio G, Bizzaro N	Autoantibody profiling of patients with primary biliary cirrhosis using a multiplexed line-blot assay	Clin Chim Acta. 438:135-8.	2015	2,764	First and corresponding authors from other institutions
Wilson's Disease	Iorio R, Ranucci G.	Wilson disease: a matter of copper, but also of zinc.	J Pediatr Gastroenterol Nutr., 60, 423-424	2015	2,4	AOU FEDERICO II NAPOLI
Wilson's Disease	Ranucci G, Di Dato F, Leone F, Vajro P, Spagnuolo MI, Iorio R.	Penicillamine-induced Elastosis Perforans Serpiginosa in Wilson Disease: Is Useful Switching to Zinc?	J Pediatr Gastroenterol Nutr., 64:e72-e73	2017	2,4	
Wilson's Disease	Ranucci G, Socha P, Iorio R.	Wilson disease: what is still unclear in pediatric patients?	Clin Res Hepatol Gastroenterol., 38, 268-272	2014	1,87	
Wilson's Disease	Ranucci G, Di Dato F, Spagnuolo MI, Vajro P, Iorio R.	Zinc monotherapy is effective in Wilson's disease patients with mild liver disease diagnosed in childhood: a retrospective study.	Orphanet J Rare Dis., 25, 41	2014	6	

Wilson's Disease	Dubbioso R, Ranucci G, Esposito M, Di Dato F, Topa A, Quarantelli M, Matarazzo M, Santoro L, Manganelli F, Iorio R.	Subclinical neurological involvement does not develop if Wilson's disease is treated early.	Parkinsonism Relat Disord, 24, 15-9	2016	5,2	
Wilson's Disease	Iorio R, Ranucci G.	Aberrance of Serum Zinc and Free Copper Level in Wilson Disease.	J Pediatr Gastroenterol Nutr., 62:e46	2016	3,2	
Shwachman-Diamond syndrome	Veropalumbo C, Campanozzi A, De Gregorio F, Correra A, Raia V, Vajro P.	Shwachman-Diamond syndrome with autoimmune-like liver disease and enteropathy mimicking celiac disease.	Clin Res Hepatol Gastroenterol., 39, e1-4	2015	1,87	Università Salerno
Liver diseases, general	Vajro P, Ferrante L, Lenta S, Mandato C, Persico M.	Management of adults with paediatric-onset chronic liver disease: strategic issues for transition care.	Dig Liver Dis. 2014 Apr;46(4):295-301	2014	2,7	
Wilson's Disease	Loudianos G, Zappu A, Lepori MB, Dessì V, Mameli E, Orrù S, Podda RA, De Virgiliis S.	Acute Liver Failure Because of Wilson Disease With Overlapping Autoimmune Hepatitis Features: The Coexistence of Two Diseases?	J Pediatr Gastroenterol Nutr., 63, e23-e24	2016	2,4	OSPEDALE MICROCITEMIE CAGLIARI
Wilson's Disease	Loudianos G, Lepori MB, Mameli E, Dessì V, Zappu A.	Wilson's disease.	Pril (Makedon Akad Nauk Umet Odd Med Nauki). 2014;35(1):93-8.	2014	1,1	
Wilson's Disease	Mameli E, Lepori MB, Chiappe F, Ranucci G, Di Dato F, Iorio R, Loudianos G.	Wilson's disease caused by alternative splicing and Alu exonization due to a homozygous 3039-bp deletion spanning from intron 1 to exon 2 of the ATP7B gene.	Gene, 15:276-279	2015	2,13	
Biliary Atresia	Sciveres M, Milazzo MP, Maggiore G.	A scoring system for biliary atresia: is this the right one?	J Hepatol, 62, 985-986	2015	11,33	ISMETT Palermo

Liver transplant	de Ville de Goyet J, Meyers RL, Tiao GM, Morland B.	Beyond the Milan criteria for liver transplantation in children with hepatic tumours.	Lancet Gastroenterol Hepatol. ,7, 456-462.	2017	10,2	
Liver diseases, general	Giordano P, Francavilla M, Buonamico P, Suppressa P, Lastella P, Sangerardi M, Miniello VL, Scardapane A, Lenato GM, Sabbà C.	Hepatic angiodynamic profile in paediatric patients with hereditary haemorrhagic telangiectasia type 1 and type 2.	Vasa. 2017 May;46(3):195-202.	2017	0,6	AO Bari
Autoimmune hepatitis	Praticò AD, Salafia S, Praticò ER, Franzonello C, Barone P, Leonardi S.	Autoimmune hepatitis: clinical findings in six pediatric patients.	Minerva Pediatr. 2016 Dec;68(6):503-505.	2016	0,53	AO Catania

Trial clinici farmacologici

Lo stato dell'arte sullo sviluppo dei farmaci per malattie rare del fegato in età pediatrica con la partecipazione alle sperimentazioni di gruppi italiani
L'epatologia pediatrica è una branca di nicchia della pediatria che copre malattie per lo più sono rare. Negli ultimi anni i ricercatori Italiani hanno preso parte a *trial* clinici per lo più coordinati in ambito europeo. Di seguito l'elenco dei trial in corso in Italia.

1	2	3		4		5	6	7		8
Nome MR	***Sponsor***	***Partners***		**Ruolo**		**Denominazione del *trial***	**Farmaco**	**Durata**		**Note**
		I	**N**	**C**	**P**			**C**	**T (anno)**	
Wilson Disease	Wilson Therapeutics AB	X	X		X	Efficacy and Safety Study of WTX101 in Wilson Disease Patients (WTX101-301)	WTX101	X		-
Wilson Disease	Univar BV	X	X		X	Study to Assess Long-Term Outcomes of Trientine in Wilson Disease Patients Withdrawn From Therapy With d-Penicillamine	Trientine	X		NCT02426905
LAL-deficiency	Alexion Pharmaceuticals	X	X		X	A Multicenter Study of SBC-102 (Sebelipase Alfa) in Patients With Lysosomal Acid Lipase Deficiency/ ARISE (Acid Lipase Replacement Investigating Safety and Efficacy)	Sebelipase Alfa	X		NCT01757184
Crigler-Najjar Syndrome	Promethera Biosciences	X	X		X	Safety Study of HepaStem for the Treatment of Crigler-Najjar Syndrome (CN) (HEP001)	HepaSTem	X		NCT01765283
Familial Cholestasis	Shire	X	X		X	A Long-Term, Open-Label Study of LUM001 With a Double-Blind, Placebo Controlled, Randomized Drug Withdrawal Period to Evaluate Safety and Efficacy in Children With Alagille Syndrome (ICONIC)	LUM001	X		NCT02160782
Familial Cholestasis		X	X		X	Meta-analysis of individual patient data of PFIC before and after surgery (bile diversion or liver transplantation)	biliary diversion	X		H7032 NAPPED Study
Genetic Cholestasis	Sahlgrenska University Hospital, Sweden	X	X		X	IBAT Inhibitor A4250 for Cholestatic Pruritus	A4250	X		NCT02360852

Partecipazione Nazionale a ERN

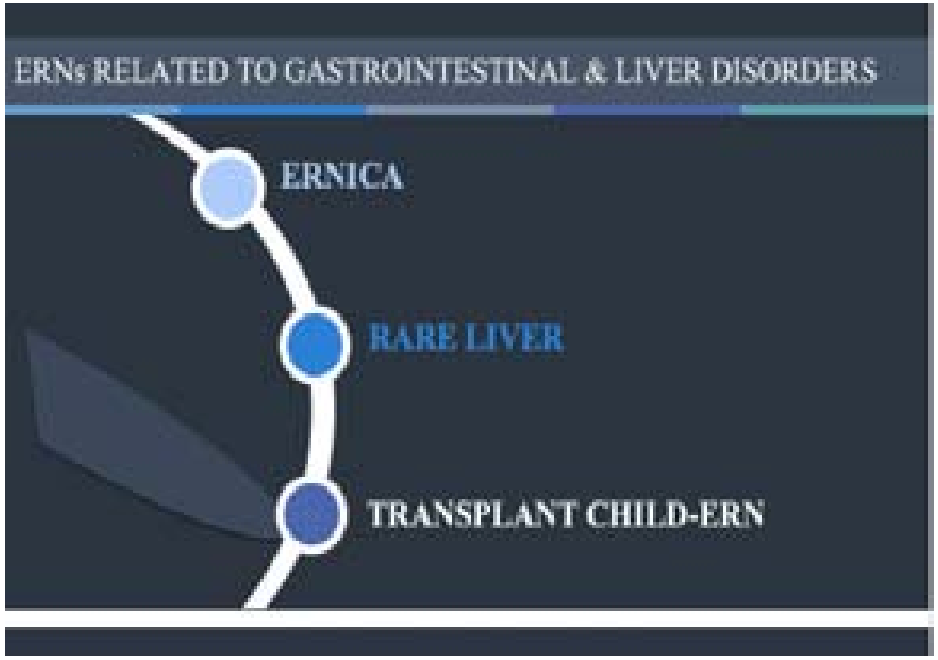
La ricognizione dei centri pediatrici che sono stati accettati nella rete europea di riferimento (ERN) per le malattie epatiche in età pediatrica

A livello nazionale da oltre 25 anni la SIGENP (Società di Gastroenterologia Epatologia e Nutrizione Pediatrica), riunisce medici, ricercatori e operatori sanitari che si dedicano allo studio e alla cura delle malattie di fegato nel bambino.

Tra gli obiettivi della Società ci sono: la promozione di studi di fisiopatologia dell'intestino, del fegato, del pancreas e di nutrizione clinica in età pediatrica, con particolare attenzione agli aspetti multidisciplinar; il supporto alla ricerca e l'educazione sulle cause, sulla prevenzione e sul trattamento delle malattie dell'apparato digerente e del fegato; lo sviluppo di relazioni scientifiche con le altre società italiane e internazionali e le attività di ricerca in gastroenterologia, epatologia e nutrizione pediatrica; la cooperazione scientifica con l'industria al fine di facilitare il raggiungimento degli scopi societari.

In ambito delle malattie rare l'obiettivo principale della SIGENP è il miglioramento della cooperazione tra gli esperiti Italiani e la facilitazione degli studi multicentrici in questo ambito. Inoltre la SIGENP si sta impegnando per migliorare la qualità dell'assistenza nelle malattie rare gastroeneterologiche ed epatologiche tramite l'elaborazione e l'applicazione di protocolli comuni nazionali e internazionali, la divulgazione della conoscenza in questo campo, la formazione degli operatori e la collaborazione con le associazioni dei genitori.

Le Reti Europee con interesse gastroenterologico ed epatologico sono 3 .



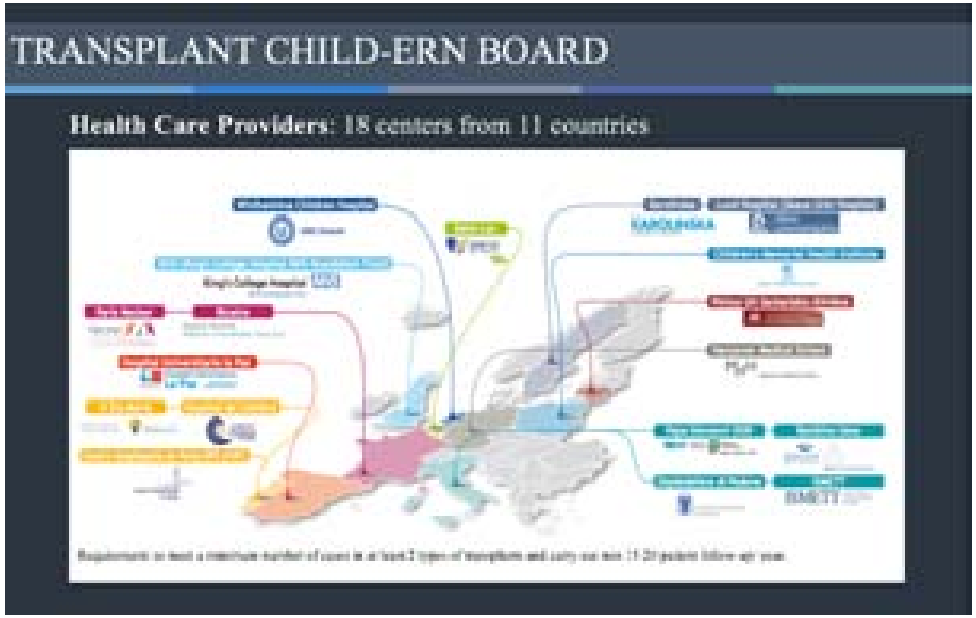
La rete di riferimento europea per le malattie rare del fegato (**ERN Rare Liver)** è coordinata dalla David Jones (The Newcastle upon Tyne Hospitals NHS Foundation Trust, United Kingdom) per garantire l'accesso a tutti i pazienti pediatrici con patologie epatiche a Centri altamente specializzati nella gestione e cura di queste patologie. La malattie d'interesse pediatrico incluse al momento nella rete fegato sono: il deficit di alfa1-antitripsina, la malattia di Wilson, le malattie cistiche del fegato, l'atresia delle vie biliari, le colestasi genetiche, le malattie vascolari del fegato, l'epatite autoimmune, la colangite sclerosante.

Questa rete prevede, mediante la collaborazione tra i diversi Centri Europei con specifiche e comprovate competenze, di garantire uniformità di cure e ridurre le disuguaglianze nell'erogazione dei servizi tra i diversi stati europei. Questa rete affronta le malattie distinguendole in tre ambiti: malattie patiche autoimmuni (coordinate da Ansgar Lohse, Deidre Kelly-referente pediatrcio), le malattie metaboliche e l'atresia delle vie biliari (coordinate da Ekkehard Strum, anche referente pediatrico) e le malattie strutturali del fegato (coordinate dal Joost Drenth, Dott. Ssa Valerie Mc Lin referente pediatrico). La rete ha già incorporato nelle sue attività 5 registri di malattie. Questa rete unendo le cure in età pediatrica con quelle dell'età adulta dovrebbe svolgere un ruolo fondamentale soprattutto nella gestione dei problemi legati alla transizione. L'ERN-Rare liver ha tra gli obiettivi l'implementazione delle linee guida sulle cure in collaborazione con l'EASL e l'ESPGHAN. La rete include 28 Centri Membri di 11 diverse Nazioni

Europee. I Centri Italiani che hanno ottenuto l'endorsement ministeriale sono 7 e rappresentano alcuni dei principali Centri SIGENP che gestiscono i pazienti pediatrici italiani affetti da patologia del fegato.

Infine ben 4 centri di Epatologia pediatrica sono coinvolti nella rete Europea sui trapianti (**ERN Transplant Child)** ed in particolare per la sezione epatotrapianto.

Centers Member ERN liver	**Minister of Health Endorsed Center**
AOU Padova (pediatric+adult)	AOU Federico II Napoli (pediatric)
San Paolo e San Carlo Milano (adult)	IRCCS Ca' Granda Milano (pediatric)
San Gerardo Monza (adult)	ISMETT Palermo (pediatric+adult)
	Città della salute e della scienza Torino (pediatric+adult)
	Policlinico Bari (adult)
	Bambino Gesù Roma (pediatric)
	Ospedale Sant'Orsola Malpighi Bologna (adult)



Per quanto concerne l'ERN sulle patologie gastrointestinali rare incluse nella rete ERNICA, la partecipazione dell'Italia è stata più scarsa. Probabilmente la nomenclatura stessa della rete (malformazioni congenite intestinali) ha sfavorito l'adesione nella prima fase.

ERNICA's BOARD

- **Network Coordinator:**
  René Wijnen (Rotterdam)
- **Deputy Coordinator:**
  Tomas Wester (Stockholm)
- **Health Care Providers:**
  20 centers from 10 countries

COUNTRY	HEALTH CARE PROVIDERS	
BELGIUM	1	Leuven
DENMARK	2	Copenhagen, Odense
FINLAND	1	Helsinki
FRANCE	6	Bicetre, Beaujon, Necker, Debré Lille, Marseille
GERMANY	2	Hannover, Mannheim
ITALY	1	Padova
NORWAY	1	Oslo
SWEDEN	1	Stockholm
NETHERLANDS	4	Amsterdam, Nijmegen, Rotterdam, Utrecht
UK	1	GOSH-London

Malattie genetiche pediatriche e sindromi malformative

A cura della Società Italiana di di Malattie Genetiche Pediatriche e Disabilità Congenite - SIMGePeD
(Presidente*: Andrea Bartuli*)

Daniela Melis
Francesca Rocchi
Giuseppe Zampino

Pubblicazioni

Società Italiana Malattie Genetiche Pediatriche e Disabilità
SIMGePeD

Numero totale dei lavori è 138

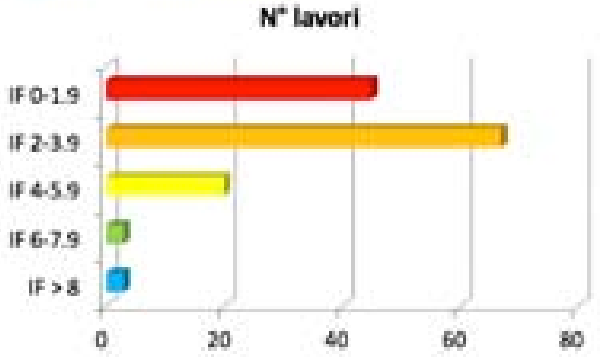
Numero medio dei lavori per socio è 4,6

1 lavoro:	8 soci
2 lavori:	5 soci
3 lavori:	5 soci
4 lavori:	2 soci
5 lavori:	1 soci
6 lavori:	3 soci
8 lavori:	2 soci
10 lavori:	1 soci
11 lavori:	1 soci
13 lavori:	1 soci
23 lavori:	1 soci

Società Italiana Malattie Genetiche Pediatriche e Disabilità
SIMGePeD

IF medio: 3.857

Range IF: 0.7 (arch ital biol) – 20 (BMJ)



Developmental Neurology Division, Carlo Besta Neurological Institute, I.R.C.C.S. Foundation, Milan, Italy

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Dysmorphic syndromes	Alfei E, Raviglione F, Franceschetti S, D'Arrigo S, Milani D, Selicorni A, Riva D, Zuffardi O, Pantaleoni C, Binelli S.	Seizures and EEG features in 74 patients with genetic-dysmorphic syndromes	Am J Med Genet A. 2014 Dec;164A(12):3154-61. doi: 10.1002/ajmg.a.36746. Epub 2014 Sep 24. PubMed PMID: 25257908.	2014	2.3	

UOC Malattie Rare e Genetica Medica/Dipartimento Pediatrico Universitario Ospedaliero/ Ospedale Pediatrico Bambino Gesù IRCCS

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Down syndrome	Diletta Valentini, M.D.; Anna Alisi; Chiara Di Camillo, MD; Maria Rita Sartorelli, MD; Annalisa Crudele; Andrea Bartuli, MD; Valerio Nobili, MD, PhD; Alberto Villani, MD, PhD	Non-alcoholic fatty liver disease in Italian children with Down syndrome: prevalence and correlation with obesity-related features The Journal of Pediatrics in press	The Journal of Pediatrics in press	2017	3.9	
Williams syndrome	De Lorenzo F, Macchiaiolo M, Carlevaris CM, Bartuli A.	The work experience of a patient affected by Williams Syndrome: a pilot project at the Bambino Gesù Children's Hospital.	Orphanet J Rare Dis. 2017 May 31;12(1):107.	2017	3.3	

Lipoprotein Lipase Deficiency	Buonuomo PS, Malamisura M, Macchiaiolo M, Rana I, Gonfiantini MV, Mastrogiorgio G, Bartuli A.	Eruptive Xanthomas in Lipoprotein Lipase Deficiency. J Pediatr. 2017 May 18. pii: S0022-3476(17)30515-2	J Pediatr. 2017 May 18. pii: S0022-3476(17)30515-2	2017	3.9	
Hypercholesterolemia	Buonuomo PS, Iughetti L, Pisciotta L, Rabacchi C, Papadia F, Bruzzi P, Tummolo A, Bartuli A, Cortese C, Bertolini S, Calandra S.	Timely diagnosis of sitosterolemia by next generation sequencing in two children with severe hypercholesterolemia.	Atherosclerosis. 2017 May 4;262:71-77.	2017	3.9	
Down syndrome	Buonuomo PS, Bartuli A, Mastrogiorgio G, Vittucci A, Di Camillo C, Bianchi S, Pires Marafon D, Villani A, Valentini D.	Lipid profiles in a large cohort of Italian children with Down syndrome.	Eur J Med Genet. 2016 Aug;59(8):392-5	2016	1	
Hypertriglyceridemia	Buonuomo PS, Bartuli A, Rabacchi C, Bertolini S, Calandra S.	A 3-day-old neonate with severe hypertriglyceridemia from novel mutations of the GPIHBP1 gene.	J Clin Lipidol. 2015 Mar-Apr;9(2):265-70.	2015	4.9	
Bazex-Duprè-Christol syndrome	Gonfiantini MV, Armando M, Pucciarini ML, Macchiaiolo M, Buonuomo PS, Diociaiuti A, Lepri FR, Sirleto P, Vicari S, Bartuli A.	Borderline cognitive level in a family with Bazex-Dupré-Christol syndrome.	Am J Med Genet A. 2015 Jul;167(7):1637-43.	2015	2	
Erythropoietic protoporphyria	Buonuomo PS, Macchiaiolo M, Gonfiantini MV, Biolcati G, Pitisci A, Villani A, Bartuli A	Erythropoietic protoporphyria in a boy.	Arch Dis Child. 2015 Jan;100(1):7. doi: 10.1136/archdischild-2014-307082	2015	3.2	
Stuve Wiedemann syndrome	Buonuomo PS, Macchiaiolo M, Cambiaso P, Rana I, Digilio MC, Bartuli A.	Long-term follow-up in Stuve-Wiedemann syndrome: a case report with articular involvement.	Clin Dysmorphol. 2014 Apr;23(2):45-6.	2014	1	
Thricho-rhino-phalangeal syndrome	Macchiaiolo M, Mennini M, Digilio MC, Buonuomo PS, Lepri FR, Gnazzo M, Grandin A, Angioni A, Bartuli A.	Thricho-rhino-phalangeal syndrome and severe osteoporosis: a rare association or a feature? An effective therapeutic approach with bisphosphonates.	Am J Med Genet A. 2014 Mar;164A(3):760-3.	2014	2	

Hypercholesterolemia	Macchiaiolo M, Buonuomo PS, Valente P, Rana I, Lepri FR, Gonfiantini MV, Bartuli A.	Corneal arcus as first sign of familial hypercholesterolemia	J Pediatr. 2014 Mar;164(3):670. doi: 10.1016/j.jpeds.2013.10.045.	2014	3.9	
Klippel-Feil syndrome	Buonuomo PS, Macchiaiolo M, Colafati GS, Rana I, Tomà P, Gonfiantini MV, Bartuli A.	Persistent neck pain in a girl: Klippel-Feil syndrome.	Arch Dis Child. 2014 Mar;99(3):290-1.	2014	3.2	

Medical Genetics Unit, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Milan, Italy

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Microriarrangiamenti cromosomici	Ronzoni L, Novelli A, Brisighelli G, Peron A, Triulzi F, Bianchi V, Leva E, Bedeschi MF	2q33.1q34 Deletion in a Girl with Brain Anomalies and Anorectal Malformation.	Cytogenet Genome Res. 2016;150(1):23-28.	2016	1.4	
Moebius syndrome	Picciolini O, Porro M, Cattaneo E, Castelletti S, Masera G, Mosca F, Bedeschi MF.	Moebius syndrome: clinical features, diagnosis, management and early intervention.	Ital J Pediatr. 2016 Jun 3;42(1):56. doi: 10.1186/s13052-016-0256-5.	2016	1.6	
Microriarrangiamenti cromosomici	Silipigni R, Cattaneo E, Baccarin M, Fumagalli M, Bedeschi MF	Rare interstitial deletion of chromosome 2p11.2p12. Report of a new patient with developmental delay and unusual clinical features	Eur J Med Genet. 2016 Jan;59(1):39-42.	2016	2.1	
Microriarrangiamenti cromosomici	Ronzoni L, Peron A, Bianchi V, Baccarin M, Guerneri S, Silipigni R, Lalatta F, Bedeschi MF.	Molecular cytogenetic characterization of a 2q35-q37 duplication and a 4q35.1-q35.2 deletion in two cousins: a genotype-phenotype analysis.	Am J Med Genet A. 2015 Jul;167(7):1551-9.	2015	2.3	
Rubinstein Taybi syndrome	Bedeschi MF, Crippa BL, Colombo L, Guez S, Cerruti M, Fogliani R, Gervasini C, Lalatta F.	Unusual prenatal presentation of Rubinstein-Taybi syndrome: a case report.	Am J Med Genet A. 2014 Oct;164A(10):2663-6.	2014	2.3	
Ehlers Danlos syndrome	Bedeschi MF, Bonarrigo F, Manzoni F, Milani D, Piemontese MR, Guez	Ehlers-Danlos syndrome versus cleidocranial dysplasia.	Ital J Pediatr. 2014 May 24;40:49.	2014	1.6	

	S, Esposito S					

Clinica pediatrica II, Ospedale Pediatrico Microcitemico A.Cao, AO Brotzu, Cagliari

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Spinocerebellar ataxia	Borroni B, Di Gregorio E, Orsi L, Vaula G, Costanzi C, Tempia F, Mitro N, Caruso D, Manes M, Pinessi L, Padovani A, Brusco A, Boccone L.	Clinical and neuroradiological features of spinocerebellar ataxia 38 (SCA38).	Parkinsonism Relat Disord. 2016 Jul;28:80-6.	2016	3.8	
Jeune syndrome	Congiu P, Puligheddu M, Gioi G, Marica M, Pani C, Piga S, Marrosu F, Boccone L	Respiratory sleep disorders in Jeune syndrome: a case description.	Arch Ital Biol. 2015 Jun-Sep;153(2-3):157-61	2015	0.7	

Unit of Clinical Genetics, San Camillo-Forlanini General Hospital, Rome, Italy

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Ehlers Danlos Syndrome	Ritelli M, Morlino S, Giacopuzzi E, Bernardini L, Torres B, Santoro G, Ravasio V, Chiarelli N, D'Angelantonio D, Novelli A, Grammatico P, Colombi M, Castori M.	A recognizable systemic connective tissue disorder with polyvalvular heart dystrophy and dysmorphism associated with TAB2 mutations.	Clin Genet. 2017 Apr 6. doi: 10.1111/cge.13032.	2017	3.3	

Ehlers Danlos Syndrome	Morlino S, Dordoni C, Sperduti I, Venturini M, Celletti C, Camerota F, Colombi M, Castori M	Refining patterns of joint hypermobility, habitus, and orthopedic traits in joint hypermobility syndrome and Ehlers-Danlos syndrome, hypermobility type.	Am J Med Genet A. 2017 Apr;173(4):914-929.	2017	2.3	
Ehlers Danlos Syndrome	Castori M, Tinkle B, Levy H, Grahame R, Malfait F, Hakim A.	A framework for the classification of joint hypermobility and related conditions.	Am J Med Genet C Semin Med Genet. 2017 Mar;175(1):148-157.	2017	4.5	
Ehlers Danlos Syndrome	Ritelli M, Morlino S, Giacopuzzi E, Carini G, Cinquina V, Chiarelli N, Majore S, Colombi M, Castori M.	Ehlers-Danlos syndrome with lethal cardiac valvular dystrophy in males carrying a novel splice mutation in FLNA.	Am J Med Genet A. 2017 Jan;173(1):169-176.	2017	2.3	
palmoplantar keratoderma	Castori M, Morlino S, Sana ME, Paradisi M, Tadini G, Angioni A, Malacarne M, Grammatico P, Iascone M, Forzano F.	Clinical and molecular characterization of two patients with palmoplantar keratoderma-congenital alopecia syndrome type 2.	Clin Exp Dermatol. 2016 Aug;41(6):632-5.		1.6	
Ehlers Danlos Syndrome	Castori M.	Ehlers-Danlos syndrome(s) mimicking child abuse: Is there an impact on clinical practice?	Am J Med Genet A. 2016 Jul;170(7):1947	2016	2.3	
Mosaicism	Castori M, Tadini G.	Discoveries and controversies in cutaneous mosaicism.	G Ital Dermatol Venereol. 2016 Jun;151(3):251-65	2016	1.3	
Aneuploidie	Castori M, Servadei F, Laino L, Pascolini G, Fabbri R, Cifani AE, Sforzolini GS, Silvestri E, Grammatico P	Axial skeletogenesis in human autosomal aneuploidies: A radiographic study of 145 second trimester fetuses.	Am J Med Genet A. 2016 Mar;170(3):676-87.	2016	2.3	
Campomelic dysplasia	Castori M, Bottillo I, Morlino S, Barone C, Cascone P; Pediatric Craniofacial Malformation (PECRAM) Study Group., Grammatico P, Laino L.	Variability in a three-generation family with Pierre Robin sequence, acampomelic campomelic dysplasia, and intellectual disability due to a novel ~1 Mb deletion upstream of SOX9, and including KCNJ2 and KCNJ16.	Birth Defects Res A Clin Mol Teratol. 2016 Jan;106(1):61-8.	2016	1.8	

Ehlers Danlos Syndrome	Castori M.	Ehlers-Danlos syndrome(s) mimicking child abuse: Is there an impact on clinical practice?	Am J Med Genet C Semin Med Genet. 2015 Dec;169(4):289-92.	2015	4.5	
Ehlers Danlos Syndrome	Sinibaldi L, Ursini G, Castori M.	Psychopathological manifestations of joint hypermobility and joint hypermobility syndrome/ Ehlers-Danlos syndrome,hypermobility type: The link between connective tissue and psychological distress revised.	Am J Med Genet C Semin Med Genet. 2015 Mar;169C(1):97-106	2015	4.5	
Ehlers Danlos Syndrome	Castori M, Morlino S, Pascolini G, Blundo C, Grammatico P	Gastrointestinal and nutritional issues in joint hypermobility syndrome/Ehlers-Danlos syndrome, hypermobility type.	Am J Med Genet C Semin Med Genet. 2015 Mar;169C(1):54-75.	2015	4.5	
Ehlers Danlos Syndrome	Castori M, Colombi M.	Generalized joint hypermobility, joint hypermobility syndrome and Ehlers-Danlos syndrome, hypermobility type.	Am J Med Genet C Semin Med Genet. 2015 Mar;169C(1):1-5.	2015	4.5	
Cerebro dermato osseous dysplasia	Castori M, Pascolini G, Parisi V, Sana ME, Novelli A, Nürnberg P, Iascone M, Grammatico P.	Microcephaly, ectodermal dysplasia, multiple skeletal anomalies and distinctive facial appearance: delineation of cerebro-dermato-osseous-dysplasia.	Am J Med Genet A. 2015 Apr;167A(4):842-51	2015	2.3	
Ehlers Danlos Syndrome	Castori M, Morlino S, Ghibellini G, Celletti C, Camerota F, Grammatico P.	Connective tissue, Ehlers-Danlos syndrome(s), and head and cervical pain.	Am J Med Genet C Semin Med Genet. 2015 Mar;169C(1):84-96.	2015	4.5	
Ehlers Danlos Syndrome	Castori M, Dordoni C, Morlino S, Sperduti I, Ritelli M, Valiante M, Chiarelli N, Zanca A, Celletti C, Venturini M, Camerota F, Calzavara-Pinton P, Grammatico P, Colombi M.	Spectrum of mucocutaneous manifestations in 277 patients with joint hypermobility syndrome/Ehlers-Danlos syndrome, hypermobility type.	Am J Med Genet C Semin Med Genet. 2015 Mar;169C(1):43-53.	2015	4.5	

Ehlers Danlos Syndrome	Ghibellini G, Brancati F, Castori M.	Neurodevelopmental attributes of joint hypermobility syndrome/Ehlers-Danlos syndrome, hypermobility type: Update and perspectives.	Am J Med Genet C Semin Med Genet. 2015 Mar;169C(1):107-16.	2015	4.5	
Ehlers Danlos Syndrome	Castori M, Voermans NC	Neurological manifestations of Ehlers-Danlos syndrome(s): A review.	Iran J Neurol. 2014 Oct 6;13(4):190-208.	2014	1	
Ehlers Danlos Syndrome	Castori M, Dordoni C, Valiante M, Sperduti I, Ritelli M, Morlino S, Chiarelli N, Celletti C, Venturini M, Camerota F, Calzavara-Pinton P, Grammatico P, Colombi M.	Nosology and inheritance pattern(s) of joint hypermobility syndrome and Ehlers-Danlos syndrome, hypermobility type: a study of intrafamilial and interfamilial variability in 23 Italian pedigrees.	Am J Med Genet A. 2014 Dec;164A(12):3010-20.	2014	2.3	
Nager syndrome	Castori M, Bottillo I, D'Angelantonio D, Morlino S, De Bernardo C, Scassellati Sforzolini G, Silvestri E, Grammatico P.	A 22-Week-Old Fetus with Nager Syndrome and Congenital Diaphragmatic Hernia due to a Novel SF3B4 Mutation.	Mol Syndromol. 2014 Aug;5(5):241-4.		1	
Microriarrandiamenti	Castori M, Bottillo I, Laino L, Morlino S, Grammatico B, Grammatico P	An additional patient with 3q27.3 microdeletion syndrome.	J Child Neurol. 2015 Mar;30(4):500-4.		1.4	
Ehlers Danlos Syndrome	Castori M, Morlino S, Grammatico P.	Towards a re-thinking of the clinical significance of generalized joint hypermobility, joint hypermobiity syndrome, and Ehlers-Danlos syndrome, hypermobility type.	Am J Med Genet A. 2014 Mar;164A(3):588-90.	2014	2.3	
Ehlers Danlos Syndrome	Castori M, Morlino S, Ritelli M, Brancati F, De Bernardo C, Colombi M, Grammatico P.	Late diagnosis of lateral meningocele syndrome in a 55-year-old woman with symptoms of joint instability and chronic musculoskeletal pain.	Am J Med Genet A. 2014 Feb;164A(2):528-34	2014	2.3	

Department of Pediatrics, "Sapienza", University of Rome, Rome, Italy

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Osteogenesis imperfecta	D'Eufemia P, Finocchiaro R, Zambrano A, Lodato V, Celli L, Finocchiaro S, Persiani P, Turchetti A, Celli M.	Serum creatine kinase isoenzymes in children with osteogenesis imperfecta.	Osteoporos Int. 2017 Jan;28(1):339-346.	2017	3.4	
Osteogeneisis imperfecta	Persiani P, Graci J, de Cristo C, Noia G, Villani C, Celli M.	Association between spondylolisthesis and L5 fracture in patients with Osteogenesis Imperfecta.	Eur Spine J. 2015 Jan 1.	2015	2.1	

Department of Medical and Surgical Sciences (DIMEC), Neonatology Unit, St. Orsola-Malpighi Polyclinic, University of Bologna, Bologna, Italy

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Down syndrome	Pelleri MC, Cicchini E, Locatelli C, Vitale L, Caracausi M, Piovesan A, Rocca A, Poletti G, Seri M, Strippoli P, Cocchi G.	Systematic reanalysis of partial trisomy 21 cases with or without Down syndrome suggests a small region on 21q22.13 as critical to the phenotype.	Hum Mol Genet. 2016 Jun 15;25(12):2525-2538.	2016	6.4	

Department of Sciences for Health Promotion and Mother and Child Care, Neonatal Intensive Care Unit, AOUP, University of Palermo, Palermo, Italy

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Down syndrome	La Placa S, Pinello G, Schierz IAM, Giuffrè M, Corsello G.	Coronary Artery Fistula in Down Syndrome: A Hidden Association.	J Ultrasound Med. 2017 Jun;36(6):1282-1283.	2017	1.544	
Congenital CMV	Colomba C, Giuffrè M, La Placa S, Cascio A, Trizzino M, De Grazia S, Corsello G.	Congenital cytomegalovirus related intestinal malrotation: a case report.	Ital J Pediatr. 2016 Dec 7;42(1):105.	2016	1.614	
Gastrointestinal malformation	Schierz IA, Pinello G, Giuffrè M, La Placa S, Piro E, Corsello G	Congenital heart defects in newborns with apparently isolated single gastrointestinal malformation: A retrospective study.	Early Hum Dev. 2016 Dec;103:43-47.	2016	1.913	
	Opitz JM, Pavone L, Corsello G.	The power of stories in Pediatrics and Genetics.	Ital J Pediatr. 2016 Apr 5;42:35.	2016	1.614	
Microriarrangiamenti	Corsello G, Salzano E, Vecchio D, Antona V, Grasso M, Malacarne M, Carella M, Palumbo P, Piro E, Giuffrè M.	Paternal uniparental disomy chromosome 14-like syndrome due a maternal de novo 160 kb deletion at the 14q32.2 region not encompassing the IG- and the MEG3-DMRs: Patient report and genotype-phenotype correlation.	Am J Med Genet A. 2015 Dec;167A(12):3130-8.	2015	2.082	
Neonatal Screning	Burlina AB, Corsello G. .	Survey of Italian pediatricians' perspectives and knowledge about neonatal screening.	Ital J Pediatr. 2015 May 29;41:41.	2015	1.614	
Microriarrangiamenti	Piccione M, Salzano E, Vecchio D, Ferrara D, Malacarne M, Pierluigi M, Ferrara I, Corsello G.	4p16.1-p15.31 duplication and 4p terminal deletion in a 3-years old Chinese girl: Array-CGH, genotype-phenotype and neurological characterization.	Eur J Paediatr Neurol. 2015 Jul;19(4):477-83.	2015	1.923	

PTEN	Piccione M, Fragapane T, Antona V, Giachino D, Cupido F, Corsello G.	Response to Stanich et al.: Correspondence regarding-PTEN hamartoma tumor syndromes in childhood-Description of two cases and a proposal for follow-up protocol.	Am J Med Genet A. 2014 Jul;164A(7):1871.	2014	2.159	
Toxoplasmosi	Puccio G, Cajozzo C, Canduscio LA, Cino L, Romano A, Schimmenti MG, Giuffrè M, Corsello G.	Epidemiology of Toxoplasma and CMV serology and of GBS colonization in pregnancy and neonatal outcome in a Sicilian population.	Ital J Pediatr. 2014 Feb 22;40:23.	2014	1.523	
Microriarrangiamenti	Palumbo P, Antona V, Palumbo O, Piccione M, Nardello R, Fontana A, Carella M, Corsello G.	Variable phenotype in 17q12 microdeletions: clinical and molecular characterization of a new case.	Gene. 2014 Apr 1;538(2):373-8.	2014	2.138	

Developmental Neurology Division, IRCCS Fondazione Istituto Neurologico C. Besta, Milan, Italy

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
CGH array	D'Arrigo S, Gavazzi F, Alfei E, Zuffardi O, Montomoli C, Corso B, Buzzi E, Sciacca FL, Bulgheroni S, Riva D, Pantaleoni C.	The Diagnostic Yield of Array Comparative Genomic Hybridization Is High Regardless of Severity of Intellectual Disability/Developmental Delay in Children.	J Child Neurol. 2016 May;31(6):691-9.	2016	1.4	Presente già in Pantaleoni
	Willemsen MA, D'Arrigo S.	Little folks, little myelin, and little teeth.	Neurology. 2014 Nov 18;83(21):1884-5.	2014	8.2	

Pediatrician Systematic, AORN Santobono-Pausilipon, Napoli, Italy

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Fraser syndrome	De Bernardo G, Giordano M, Di Toro A, Sordino D, De Brasi D.	Prenatal diagnosis of Fraser syndrome: a matter of life or death?	Ital J Pediatr. 2015 Nov 9;41:86. doi: 10.1186/s13052-015-0195-6.	2015	1.6	

Genetics and Rare Diseases Research Division, Bambino Gesù Children's Hospital, IRCCS, Rome, Italy

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Kabuki	Dentici ML, Barresi S, Niceta M, Pantaleoni F, Pizzi S, Dallapiccola B, Tartaglia M, Digilio MC.	Clinical spectrum of Kabuki-like syndrome caused by HNRNPK haploinsufficiency.	Clin Genet. 2017 Apr 4. doi: 10.1111/cge.13029	2017	3,3	
Congenital Heart Defects	Digilio MC, Marino B.	What Is New in Genetics of Congenital Heart Defects?	Front Pediatr. 2016 Dec 1;4:120.	2016	2,2	
Noonan	Calcagni G, Baban A, Lepri FR, Marino B, Tartaglia M, Digilio MC.	Congenital heart defects in Noonan syndrome and RIT1 mutation.	Genet Med. 2016 Dec;18(12):1320.	2016	8.2	
Noonan	Calcagni G, Baban A, De Luca E, Leonardi B, Pongiglione G, Digilio MC.	Coronary artery ectasia in Noonan syndrome: Report of an individual with SOS1 mutation and literature review.	Am J Med Genet A. 2016 Mar;170(3):665-9.	2016	2.3	
22q11.2	Digilio MC, Versacci P, Bernardini L, Novelli A, Marino B, Dallapiccola B.	Left ventricular non compaction with aortic valve anomalies: A recurrent feature of 22q11.2 distal deletion syndrome.	Eur J Med Genet. 2015 Aug;58(8):406-8.	2015	1.2	
Adams oliver	Digilio MC, Marino B, Baban A, Dallapiccola B.	Cardiovascular malformations in Adams-Oliver syndrome.	Am J Med Genet A. 2015 May;167A(5):1175-7.	2015	2.3	

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
CNV	Di Gregorio E, Riberi E, Belligni EF, Biamino E, Spielmann M, Ala U, Calcia A, Bagnasco I, Carli D, Gai G, Giordano M, Guala A, Keller R, Mandrile G, Arduino C, Maffè A, Naretto VG, Sirchia F, Sorasio L, Ungari S, Zonta A, Zacchetti G, Talarico F, Pappi P, Cavalieri S, Giorgio E, Mancini C, Ferrero M, Brussino A, Savin E, Gandione M, Pelle A, Giachino DF, De Marchi M, Restagno G, Provero P, Silengo MC, Grosso E, Buxbaum JD, Pasini B, De Rubeis S, Brusco A, Ferrero GB.	CNVs analysis in a cohort of isolated and syndromic DD/ID reveals novel genomic disorders, position effects and candidate disease genes.	Clin Genet. 2017 Mar 14. doi: 10.1111/cge.13009.	2017	3,982	
Beckwith Wiedemann syndrome	Mussa A, Ferrero GB.	Serum alpha-fetoprotein screening for hepatoblastoma in Beckwith-Wiedemann syndrome.	Am J Med Genet A. 2017 Mar;173(3):585-587.	2017	2,082	
Noonan Syndrome	Baldassarre G, Mussa A, Carli D, Molinatto C, Ferrero GB.	Constitutional bone impairment in Noonan syndrome.	Am J Med Genet A. 2017 Mar;173(3):692-698.	2017	2,082	
Beckwith Wiedemann syndrome	Mussa A, Molinatto C, Baldassarre G, Riberi E, Russo S, Larizza L, Riccio A, Ferrero GB.	Cancer Risk in Beckwith-Wiedemann Syndrome: A Systematic Review and Meta-Analysis Outlining a Novel (Epi)Genotype Specific Histotype Targeted Screening Protocol.	J Pediatr. 2016 Sep;176:142-149. Cited in Science, 2017, Jan 355 (6321): 122-125.	2016	3,890	
Syndromic craniosynostosis	Biamino E, Canale A, Lacilla M, Marinosci A, Dagna F, Genitori L, Peretta P, Silengo M, Albera R, Ferrero GB.	Prevention and management of hearing loss in syndromic craniosynostosis: A case series.	Int J Pediatr Otorhinolaryngol. 2016 Jun;85:95-8.	2016	1,125	

Beckwith Wiedemann syndrome	Mussa A, Russo S, de Crescenzo A, Freschi A, Calzari L, Maitz S, Macchiaiolo M, Molinatto C, Baldassarre G, Mariani M, Tarani L, Bedeschi MF, Milani D, Melis D, Bartuli A, Cubellis MV, Selicorni A, Silengo MC, Larizza L, Riccio A, Ferrero GB.	Fetal growth patterns in Beckwith-Wiedemann syndrome.	Clin Genet. 2016 Jul;90(1):21-7.	2016	3,982	
Beckwith Wiedemann syndrome	Mussa A, Di Candia S, Russo S, Catania S, De Pellegrin M, Di Luzio L, Ferrari M, Tortora C, Meazzini MC, Brusati R, Milani D, Zampino G, Montirosso R, Riccio A, Selicorni A, Cocchi G, Ferrero GB.	Recommendations of the Scientific Committee of the Italian Beckwith-Wiedemann Syndrome Association on the diagnosis, management and follow-up of the syndrome.	Eur J Med Genet. 2016 Jan;59(1):52-64.	2016	1,810	
Beckwith Wiedemann syndrome	Mussa A, Ferrero GB.	Screening Hepatoblastoma in Beckwith-Wiedemann Syndrome: A Complex Issue.	J Pediatr Hematol Oncol. 2015 Nov;37(8):627.	2015	1,146	
Beckwith Wiedemann syndrome	Mussa A, Russo S, Larizza L, Riccio A, Ferrero GB.	(Epi)genotype-phenotype correlations in Beckwith-Wiedemann syndrome: a paradigm for genomic medicine.	Clin Genet. 2015 Jul 3. doi: 10.1111/cge.12635	2015	3,982	
Beckwith Wiedemann syndrome	Mussa A, Russo S, De Crescenzo A, Freschi A, Calzari L, Maitz S, Macchiaiolo M, Molinatto C, Baldassarre G, Mariani M, Tarani L, Bedeschi MF, Milani D, Melis D, Bartuli A, Cubellis MV, Selicorni A, Cirillo Silengo M, Larizza L, Riccio A, Ferrero GB.	Epi)genotype-phenotype correlations in Beckwith-Wiedemann syndrome.	Eur J Hum Genet. 2016 Feb;24(2):183-90.	2016	4,580	
Mazzanti syndrome	Baldassarre G, Mussa A, Banaudi E, Rossi C, Tartaglia M, Silengo M, Ferrero GB.	Phenotypic variability associated with the invariant SHOC2 c.4A>G (p.Ser2Gly) missense mutation.	Am J Med Genet A. 2014 Dec;164A(12):3120-5	2014	2,159	

Beckwith Wiedemann syndrome	Mussa A, Pagliardini S, Pagliardini V, Molinatto C, Baldassarre G, Corrias A, Silengo MC, Ferrero GB.	α-Fetoprotein assay on dried blood spot for hepatoblastoma screening in children with overgrowth-cancer predisposition syndromes.	Pediatr Res. 2014 Dec;76(6):544-8.	2014	2,2314	
CNV	Biamino E, Di Gregorio E, Belligni EF, Keller R, Riberi E, Gandione M, Calcia A, Mancini C, Giorgio E, Cavalieri S, Pappi P, Talarico F, Fea AM, De Rubeis S,Cirillo Silengo M, Ferrero GB, Brusco A.	A novel 3q29 deletion associated with autism, intellectual disability, psychiatric disorders, and obesity.	Am j Med Genet B Neuropsychiatr Genet 2016 Mar, 171B(2) 290-9	2016	3,391	

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
22q11 del	Ficcadenti A, Zallocco F, Neri R, Giovannini L, Tirabassi G, Balercia G.	Bone density assessment in a cohort of pediatric patients affected by 22q11DS.	J Endocrinol Invest. 2015 Oct;38(10):1093-8.	2015	2.6	

Division of Pediatric, Department of Clinical Sciences, Università Politecnica delle Marche, Ospedali Riuniti, Ancona, Italy

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
CHARGE	Santoro L, Ficcadenti A, Zallocco F, Del Baldo G, Piraccini F, Gesuita R, Ceccarani P, Gabrielli O.	Cognitive-motor profile, clinical characteristics and diagnosis of CHARGE syndrome: an Italian experience.	Am J Med Genet A. 2014 Dec;164A(12):3042-51.	2014		

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Microriarrangiamenti	Fischetto R, Palumbo O, Ortolani F, Palumbo P, Leone MP, Causio FA, Pesce S, Digilio MC, Carella M, Papadia F.	Clinical and molecular characterization of a second family with the 12q14 microdeletion syndrome and review of the literature.	Am J Med Genet A. 2017 Apr 13. doi: 10.1002/ajmg.a.38253	2017	2.3	
Microriarrangiamenti	Palumbo O, Palumbo P, Leone MP, Stallone R, Palladino T, Vendemiale M, Palladino S, Papadia F, Carella M, Fischetto R	PARK2 Microduplication: Clinical and Molecular Characterization of a Further Case and Review of the Literature.	Mol Syndromol. 2016 Oct;7(5):282-286.	2016	1	

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
17p13.1 microduplication syndrome	Maini I, Ivanovski I, Iodice I, Rosato S, Pollazzon M, Mussini M, Belligni EF, Coutton C, Marinelli M, Barbieri V, Napoli M, Pascarella R, Sartori C, Madia F, Fusco C, Franchi F, Street M, Garavelli L.	Endocrinological Abnormalities Are a Main Feature of 17p13.1 Microduplication Syndrome: A New Case and Literature Review				

Mowat-Wilson syndrome	Garavelli L, Ivanovski I, Caraffi SG, Santodirocco D, Pollazzon M, Cordelli DM, Abdalla E, Accorsi P, Adam MP, Baldo C, Bayat A, Belligni E, Bonvicini F, Breckpot J, Callewaert B, Cocchi G, Cuturilo G, Devriendt K, Dinulos MB, Djuric O, Epifanio R, Faravelli F, MD, Formisano D, Giordano L, Grasso M, Grønborg S, Iodice A, Iughetti L, Lacombe D, Maggi M, Malbora B, Mammi I, Moutton S, Møller R, Muschke P, Napoli M, Pantaleoni C, Pascarella R, Pellicciari A, Poch Olive ML, Raviglione F, Rivieri F, Russo C, Savasta S, Scarano G, Selicorni A, Silengo M, Sorge G, Tarani L, Gonzaga Tone L,Toutain A, Trimouille A, Valera ET, Schrier Vergano S, Zanotta N, Zollino M, Dobyns WB, Paciorkowski AR.	Neuroimaging findings in Mowat-Wilson syndrome: a study of 54 patients.				
Myhre syndrome	Garavelli L, Maini I, Baccilieri F, Ivanovski I, Pollazzon M, Rosato S, Iughetti L, Unger S, Superti-Furga A, Tartaglia M.	Natural history and life-threatening complications in Myhre syndrome and review of the literature.				
RIN2 syndrome	Rosato S, Syx D, Ivanovski I, Pollazzon M, Santodirocco D, De Marco L, Beltrami M, Callewaert B, Garavelli L, Malfait F.	RIN2 syndrome: Expanding the clinical phenotype				

Hematologic rare diseases (TEL-AML1 (ETV6-RUNX1) fusion gene - genetic abnormality in childhood Bcell precursor acute lymphoblastic leukemia.	Ivanovski I, Garavelli L, Djurić O, Ćirović A, Škorić D, Ivanovski PI.	Mitotic crossover promotes leukemogenesis in children born with TEL-AML1 via the generation of loss of heterozygosity at 12p				
Noonan syndrome-like Disorder with loose Anagen Hair (gene SHOC2)	LGaravelli, V Cordeddu, S Errico, P Bertolini, ME Street, S Rosato, M Pollazzon , A Wischmeijer,I Ivanovski, P Daniele, E Bacchini, AA Lombardi, G Izzi, G Biasucci, C Del Rossi, D Corradi, G Cazzaniga, C Dominici, C Rossi, A De Luca, S Bernasconi, R Riccardi, E Legius, M Tartaglia	Noonan Syndrome-Like Disorder with Loose Anagen Hair: a second case with Neuroblastoma				
Noonan syndrome-like Disorder with loose Anagen Hair (gene SHOC2)	Gargano Giancarlo, Guidotti Isotta, Balestri Eleonora, Vagnarelli Federica, Rosato Simonetta, Comitini Giuseppina, Wischmeijer Anita, La Sala Giovanni Battista, Iughetti Lorenzo, Cordeddu Viviana, Rossi Cesare, Tartaglia Marco, Garavelli Livia.	Hydrops fetalis in a preterm newborn heterozygous for the c.4A>G SHOC2 mutation.	Am J Med Genet A. 2014 164A(4):1015-20	2014	IF:2,304	

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Complex children	Geremia C, De Ioris MA, Crocoli A, Adorisio O, Scrocca R, Lombardi MH, Staccioli S, Stella P, Amendola P, Ciliento G, De Peppo F, Campana A.	Totally implantable venous access devices in children with medical complexity: preliminary data from a tertiary care hospital.	J Vasc Access. 2017 May 24:0. doi: 10.5301/jva.5000727.	2017	1.5	

Neonatal Intensive Care Unit, AOUP "P. Giaccone", Department of Sciences for Health Promotion and Mother and Child Care "G. D'Alessandro", University of Palermo, Italy

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Down syndrome	La Placa S, Pinello G, Schierz IAM, Giuffrè M, Corsello G.	Coronary Artery Fistula in Down Syndrome: A Hidden Association.	J Ultrasound Med 2017; 36(6): 1282-1283.	2017	1.544	
infant of diabetic mother	Schierz IAM, Pinello G, Piro E, Giuffrè M, La Placa S, Corsello G.	Transitional hemodynamics in infants of diabetic mothers by targeted neonatal echocardiography, electrocardiography and peripheral flow study.	J Matern Fetal Neonatal Med 2017; 5: 1-8.	2017	1.674	
congenital heart defects	Schierz IAM, Piro E, Giuffrè M, Pinello G, Corsello G.	Dilated azygos arch mimicking an aortic arch anomaly during thoracic surgery.	Early Hum Dev 2017; 20;111: 20-22.	2017	1.913	
congenital heart defects	Schierz IA, Pinello G, Giuffrè M, La Placa S, Piro E, Corsello G.	Congenital heart defects in newborns with apparently isolated single gastrointestinal malformation: A retrospective study.	Early Hum Dev 2016; 103: 43-47.	2016	1.913	
microriarrangiamenti	Vecchio D, Giuffrè M.	The Coat-Hanger Angle Sign.	J Pediatr 2016; 177: 325-325.e1.	2016	3.890	

microriarrangiamenti	Corsello G, Salzano E, Vecchio D, Antona V, Grasso M, Malacarne M, Carella M, Palumbo P, Piro E, Giuffrè M.	Paternal uniparental disomy chromosome 14-like syndrome due a maternal de novo 160 kb deletion at the 14q32.2 region not encompassing the IG- and the MEG3-DMRs: Patient report and genotype-phenotype correlation.	Am J Med Genet A 2015; 167A(12): 3130-8	2015	2.082	
anomalies of penis DSD	Cimador M, Catalano P, Ortolano R, Giuffrè M.	The inconspicuous penis in children.	Nat Rev Urol 2015; 12(4): 205-15.	2015	5.957	
Pierre Robin sequence	Salerno S, Gagliardo C, Vitabile S, Militello C, La Tona G, Giuffrè M, Lo Casto A, Midiri M.	Semi-automatic volumetric segmentation of the upper airways in patients with Pierre Robin sequence.	Neuroradiol J 2014; 27(4): 487-94.	2014		

Paediatric Endocrinology Unit, Paediatric Department 'Sant'Orsola Malpighi' University Hospital, Bologna, Italy; Paediatric Endocrinology Unit, Paediatric Department 'Sant'Orsola Malpighi' University Hospital, Bologna, Italy

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Pseudohypoparathyroidism	Visconti P, Posar A, Scaduto MC, Russo A, Tamburrino F, Mazzanti L.	Neuropsychiatric phenotype in a child with pseudohypoparathyroidism	J Pediatr Neurosci. 2016 Jul-Sep;11(3):267-270.	2016	0.6	
RASopathies	Tamburrino F, Gibertoni D, Rossi C, Scarano E, Perri A, Montanari F, Fantini MP, Pession A, Tartaglia M, Mazzanti L.	Response to long-term growth hormone therapy in patients affected by RASopathies and growth hormone deficiency: Patterns of growth, puberty and final height data.	Am J Med Genet A. 2015 Nov; 167A(11):2786-94.	2015	2.3	
Cromosomopatia	D'Alberton F, Assante MT, Foresti M, Balsamo A, Bertelloni S, Dati E, Nardi L, Bacchi ML, Mazzanti L.	Quality of Life and Psychological Adjustment of Women Living with 46,XY Differences of Sex Development. J	J Sex Med. 2015 Jun;12(6):1440-9.	2015	3.2	

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Primrose syndrome	Casertano A, Fontana P, Hennekam RC, Tartaglia M, Genesio R, Dieber TB, Ortega L, Nitsch L, Melis D.	Alterations in metabolic patterns have a key role in diagnosis and progression of primrose syndrome.				
Cgh array	Cappuccio G, Vitiello F, Casertano A, Fontana P, Genesio R, Bruzzese D, Ginocchio VM, Mormile A, Nitsch L, Andria G, Melis D.	New insights in the interpretation of array-CGH: autism spectrum disorder and positive family history for intellectual disability predict the detection of pathogenic variants.				

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
DSD	Cimador M., Catalano P., Ortolano R., Giuffrè M.	The inconspicuous penis in children	Nat Rev Urol. 2015 Apr;12(4):205-215.	2015	4.84	Disorders/differences of sex development
Gastroschisis	Insinga V, Lo Verso C, Antona V, Cimador M, Ortolano R, Carta M, La Placa S, Giuffrè M, Corsello G.	*Perinatal management of gastroschisis*	Journal of Pediatric and Neonatal Individualized Medicine 2014;3(1):e030113	2014	0	

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Microriarrangiamenti	Zanzottera C, Milani D, Alfei E, Rizzo A, D'Arrigo S, Esposito S, Pantaleoni C.	ZC4H2 deletions can cause severe phenotype in female carriers.	Am J Med Genet A. 2017 May;173(5):1358-1363.	2017	2.082	
Microriarrangiamenti	Natacci F, Alfei E, Tararà L, D'Arrigo S, Zuffardi O, Gentilin B, Pantaleoni C.	Chromosome 17q21.31 duplication syndrome: Description of a new familiar case and further delineation of the clinical spectrum.	Eur J Paediatr Neurol. 2016 Jan;20(1):183-7.	2016	1.923	
Microriarrangiamenti	D'Arrigo S, Gavazzi F, Alfei E, Zuffardi O, Montomoli C, Corso B, Buzzi E, Sciacca FL, Bulgheroni S, Riva D, Pantaleoni C.	The Diagnostic Yield of Array Comparative Genomic Hybridization Is High Regardless of Severity of Intellectual Disability/Developmental Delay in Children.	J Child Neurol. 2016 May;31(6):691-9.	2016	1.434	

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Dysmorphology	Schepis C, Romano C.	Facies: the value of an old diagnostic tip in pediatric dermatology.	G Ital Dermatol Venereol. 2017 Apr 19. doi: 10.23736/S0392-0488.17.05626-7.	2017	1.014	
Down syndrome	Salemi M, Barone C, Salluzzo MG, Giambirtone M, Scillato F, Galati Rando R, Romano C, Morale MC, Ridolfo F, Romano C.	A polymorphism (rs1042522) in TP53 gene is a risk factor for Down Syndrome in Sicilian mothers.	J Matern Fetal Neonatal Med. 2016 Dec 7:1-3.	2016	1.674	
Down syndrome	Salemi M, Barone C, Morale MC, Caniglia S, Romano C, Salluzzo MG, Rando RG, Ragalmuto A, Bosco P, Romano C	Killer-specific secretory (Ksp37) gene expression in subjects with Down's syndrome	Neurol Sci. 2016 May; 37(5):793-5.	2016	1.783	

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
CHARGE	Santoro L, Ficcadenti A, Zallocco F, Del Baldo G, Piraccini F, Gesuita R, Ceccarani P, Gabrielli O.	Cognitive-motor profile, clinical characteristics and diagnosis of CHARGE syndrome: an Italian experience.	Am J Med Genet A. 2014 Dec;164A(12):3042-51.	2014	2.3	

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1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Ehlers Danlos syndrome	Lisi C, Monteleone S, Tinelli C, Rinaldi B, Di Natali G, Savasta S.	Postural analysis in a pediatric cohort of patients with Ehlers-Danlos Syndrome: a pilot study.	Minerva Pediatr. 2017 Feb 17. doi: 10.23736/S0026-4946.17.04681-3.	2017	0.532	
Paediatric clinically isolated syndromes	Trabatti C, Foiadelli T, Spartà MV, Gagliardone C, Rinaldi B, Delmonte M, Lozza A, Savasta S.	Paediatric clinically isolated syndromes: report of seven cases, differential diagnosis and literature review	Childs Nerv Syst. 2016 Jan;32(1):69-77. doi: 10.1007/s00381-015-2959-0	2016	1.080	
Ehlers Danlos syndrome	Savasta S, Verrotti A, Spartà MV, Foiadelli T, Villa MP, Parisi P.	Unilateral periventricular heterotopia and epilepsy in a girl with Ehlers-Danlos syndrome.	Epilepsy Behav Case Rep. 2015 Jun 14;4:27-9. doi: 10.1016/j.ebcr.2015.05.004	2015	-----	
Stickler syndrome	Savasta S, Salpietro V, Spartà MV, Foiadelli T, Laino D, Lobefalo L, Marseglia GL, Verrotti A.	Stickler syndrome associated with epilepsy: report of three cases-8	Eur J Pediatr. 2015 May;174(5):697-701. doi: 10.1007/s00431-015-2514-8	2015	1.791	

epilepsy	Savasta S, Budetta M, Spartà MV, Carpentieri ML, Trasimeni G, Zavras N, Villa MP, Parisi P.	Gelastic epilepsy without hypothalamic hamartoma: three additional cases.	Epilepsy Behav. 2014 Aug;37:87-90. doi: 10.1016/j.yebeh.2014.06.012	2014	2.332	
Ehlers Danlos syndrome	Verrotti A, Spartà MV, Monacelli D, Porto R, Castagnino M, Russo Raucci A, Compagno F, Viglio S, Foiadelli T, Nicita F, Grosso S, Spalice A, Chiarelli F, Marseglia G, Savasta S.	Long-term prognosis of patients with Ehlers-Danlos syndrome and epilepsy.	Epilepsia. 2014 Aug;55(8):1213-9. doi: 10.1111/epi.12699	2014	4.706	

Genetica Medica, Ospedale G. Rummo, Benevento

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Prader Willi syndrome	Fontana P, Grasso M, Acquaviva F, Gennaro E, Galli ML, Falco M, Scarano F, Scarano G, Lonardo F.	SNORD116 deletions cause Prader-Willi syndrome with a mild phenotype and macrocephaly.	Clin Genet. 2017 Mar 7. doi: 10.1111/cge.13005.	2017	3.3	
Smith Magenis syndrome	Acquaviva F, Sana ME, Della Monica M, Pinelli M, Postorivo D, Fontana P, Falco MT, Nardone AM, Lonardo F, Iascone M, Scarano G.	First evidence of Smith-Magenis syndrome in mother and daughter due to a novel RAI mutation.	Am J Med Genet A. 2017 Jan;173(1):231-238.	2017	2.3	
Prenatal test	Lonardo F, Scarano G.	The promise of non-invasive prenatal testing needs to be monitored scientifically.	BMJ. 2015 May 14;350:h2518	2015	20	
Risk Factors	Mastroiacovo P, Nilsen RM, Leoncini E, Gastaldi P, Allegri V, Boiani A, Faravelli F, Ferrazzoli F, Guala A, Madrigali V, Scarano G.	Prevalence of maternal preconception risk factors: an Italian multicenter survey.	Ital J Pediatr. 2014 Nov 23;40:91.	2014	1.6	

Genetica Clinica Pediatrica, Ospedale San Gerardo, Monza

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Baraitser Winter syndrome	Cianci P, Fazio G, Casagranda S, Spinelli M, Rizzari C, Cazzaniga G, Selicorni A.	Acute myeloid leukemia in Baraitser-Winter cerebrofrontofacial syndrome.	Am J Med Genet A. 2017 Feb;173(2):546-549.	2017	2.3	
Williams syndrome	Decimi V, Fazio G, Dell'Acqua F, Maitz S, Galbiati M, Rizzari C, Biondi A, Cazzaniga G, Selicorni A.	Williams syndrome and mature B-Leukemia: A random association?	Eur J Med Genet. 2016 Dec;59(12):634-640	2016	1.8	
Microriarrangiamenti cromosomici	Villa N, Scatigno A, Redaelli S, Conconi D, Cianci P, Farina C, Fossati C, Dalprà L, Maitz S, Selicorni A.	14q32.3-qter trisomic segment: a case report and literature review.	Mol Cytogenet. 2016 Aug 5;9:60. doi: 10.1186/s13039-016-0265-5.	2016	1.5	
Carey-Fineman-Ziter syndrome	Pasetti M, Mazzoleni F, Novelli G, Iascone M, Bozzetti A, Selicorni A.	Temporomandibular joint ankylosis as part of the clinical spectrum of Carey-Fineman-Ziter syndrome?	Am J Med Genet A. 2016 Aug;170(8):2191-5.	2016	2.3	
Cornelia de Lange syndrome	Mariani M, Decimi V, Bettini LR, Maitz S, Gervasini C, Masciadri M, Ajmone P, Kullman G, Dinelli M, Panceri R, Cereda A, Selicorni A.	Adolescents and adults affected by Cornelia de Lange syndrome: A report of 73 Italian patients.	Am J Med Genet C Semin Med Genet. 2016 Jun;172(2):206-13.	2016	4.5	
Cornelia de Lange	Cereda A, Mariani M, Rebora P, Sajeva A, Ajmone PF, Gervasini C, Russo S, Kullmann G, Valsecchi G, Selicorni A.	A new prognostic index of severity of intellectual disabilities in Cornelia de Lange syndrome.	Am J Med Genet C Semin Med Genet. 2016 Jun;172(2):179-89.	2016	4.5	
Cornelia de Lange	Moretto A, Scaravilli V, Ciceri V, Bosatra M, Giannatelli F, Ateniese B, Mariani M, Cereda A, Sosio S, Zanella A, Pesenti A, Selicorni A.	Sedation and general anesthesia for patients with Cornelia De Lange syndrome: A case series.	Am J Med Genet C Semin Med Genet. 2016 Jun;172(2):222-8.	2016	4.5	

Cornelia de Lange	Zambrelli E, Fossati C, Turner K, Taiana M, Vignoli A, Gervasini C, Russo S, Furia F, Masciadri M, Ajmone P, Kullman G, Canevini MP, Selicorni A.	Sleep disorders in Cornelia de Lange syndrome. Am J Med Genet C Semin Med Genet. 2016 Jun;172(2):214-21.	Am J Med Genet C Semin Med Genet. 2016 Jun;172(2):214-21.	2016	4.5	
Trisomy 18	Motta S, Sala D, Sala A, Cazzaniga G, Giudici G, Villa N, Biondi A, Selicorni A.	Hodgkin lymphoma in a patient with mosaic trisomy 18: First clinical observation.	Am J Med Genet A. 2016 Mar;170(3):777-80.	2016	2.3	
Cornelia de Lange	Cavalleri V, Bettini LR, Barboni C, Cereda A, Mariani M, Spinelli M, Gervasini C, Russo S, Biondi A, Jankovic M, Selicorni A.	Thrombocytopenia and Cornelia de Lange syndrome: Still an enigma?	Am J Med Genet A. 2016 Jan;170A(1):130-4.	2016	2.3	
RASopathies	Gaipa G, Bugarin C, Cianci P, Sarno J, Bonaccorso P, Biondi A, Selicorni A.	Peripheral blood cells from children with RASopathies show enhanced spontaneous olonies growth in vitro and hyperactive RAS signaling.	Blood Cancer J. 2015 Jul 17;5:e324. doi: 10.1038/bcj.2015.52.	2015	4.4	
Cornelia de Lange	Bettini LR, Locatelli L, Mariani M, Cianci P, Giussani C, Canonico F, Cereda A, Russo S, Gervasini C, Biondi A, Selicorni A.	Cervical spine malformation in cornelia de lange syndrome: a report of three patients.	Am J Med Genet A. 2014 Jun;164A(6):1520-4.	2014	2.3	

Department of Pediatrics, La Sapienza University of Rome, Italy

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Klinefelter	Pimpolari L, Liberati N, Martini M, Colloridi F, Radicioni A, Duse M, Tarani L.	Prenatal genetic counseling in Klinefelter syndrome: comments on the article by Lalatta et al. [2013] and a proposal of a new approach.	Am J Med Genet A. 2015 Feb;167A(2):450-4.	2015	2.3	

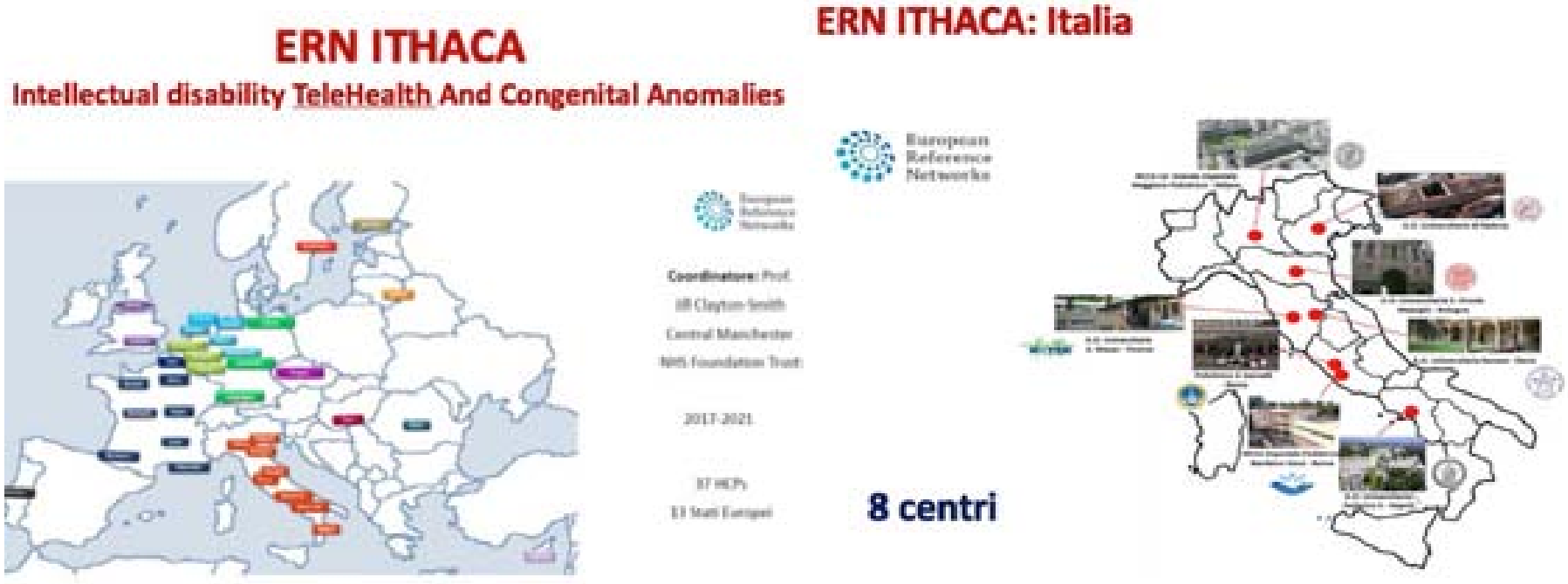
Pediatric and Infectious Disease Unit, Bambino Gesù Children's Hospital, IRCCS, Rome, Italy

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Down Syndrome	Valentini D, Alisi A, Di Camillo C, Sartorelli MR, Crudele A, Bartuli A, Nobili V, Villani A.	Non-alcoholic fatty liver disease in Italian children with Down syndrome: prevalence and correlation with obesity-related features.	Journal of Pediatrics, in press	2017	3.890	Accettato il 30 Maggio 2017
Down Syndrome	Marasco E, Farroni C, Cascioli S, Marcellini V, Scarsella M, Giorda E, Piano Mortari E, Leonardi L, Scarselli A, Valentini D, Cancrini C, Duse M, Grimsholm O, Carsetti R.	B-cell activation with CD40L or CpG measures the function of B-cell subsets and identifies specific defects in immunodeficient patients.	See comment in PubMed Commons belowEur J Immunol. 2017 Jan;47(1):131-143.	2017	4.179	
Down Syndrome	Buonuomo PS, Bartuli A, Mastrogiorgio G, Vittucci A, Di Camillo C, Bianchi S, Pires Marafon D, Villani A, Valentini D.	Lipid profiles in a large cohort of Italian children with Down syndrome.	Eur J Med Genet. 2016 Aug;59(8):392-5	2016	1.810	
Down Syndrome	Valentini D, Bianchi S, Di Camillo C, Vittucci AC, Gonfiantini MV, De Vito R, Villani A.	Fatal varicella pneumonia in an unvaccinated child with Down Syndrome: a case report.	Ital J Pediatr. 2016 Nov 17;42(1):99	2016	1.614	
Down Syndrome	Valentini D, Marcellini V, Bianchi S, Villani A, Facchini M, Donatelli I, Castrucci MR, Marasco E, Farroni C, Carsetti R.	Generation of switched memory B cells in response to vaccination in Down syndrome children and their siblings.	Vaccine. 2015 Nov 27;33(48):6689-96.	2015	3.413	
Down Syndrome	Carsetti R, Valentini D, Marcellini V, Scarsella M, Marasco E, Giustini F, Bartuli A, Villani A, Ugazio AG.	Reduced numbers of switched memory B cells with high terminal differentiation potential in Down syndrome.	Eur J Immunol. 2015 Mar;45(3):903-14	2015	4.179	

UOSA MALATTIE RARE E DIFETTI CONGENITI/ DIPARTIMENTO DONNA BAMBINO/FONDAZIONE POLICLINICO UNIVERSITARIO "A. GEMELLI"

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Wiedemann Steiner Syndrome	Stellacci E, Onesimo R, Bruselles A, Pizzi S, Battaglia D, Leoni C, Zampino G, Tartaglia M.	Congenital immunodeficiency in an individual with Wiedemann-Steiner syndrome due to a novel missense mutation in KMT2A	Am J Med Genet A. Sep;170(9):2389-93	2016	2.3	Zampino corresponding author
Auricolo-condilar syndrome	Leoni C, Gordon CT, Della Marca G, Giorgio V, Onesimo R, Perrino F, Cianfoni A, Cerchiari A, Amiel J, Zampino G	Respiratory and gastrointestinal dysfunctions associated with auriculo-condylar syndrome and a homozygous PLCB4 loss-of-function mutation.	Am J Med Genet A. Jun;170(6):1471-8.	2016	2.3	
Costello syndrome	Leoni C, Onesimo R, Giorgio V, Diamanti A, Giorgio D, Martini L, Rossodivita A, Tartaglia M, Zampino G	Understanding Growth Failure in Costello Syndrome: Increased Resting Energy Expenditure.	J Pediatr. 2016 Mar;170:322-4.	2016	3.9	
Ceroid-lipofuscinosis	Di Giacopo R, Cianetti L, Caputo V, La Torraca I, Piemonte F, Ciolfi A, Petrucci S, Carta C, Mariotti P, Leuzzi V, Valente EM, D'Amico A, Bentivoglio A, Bertini E, Tartaglia M, Zampino G.	Protracted late infantile ceroid lipofuscinosis due to TPP1 mutations: Clinical, molecular and biochemical characterization in three sibs.	J Neurol Sci. 2015 Sep 15;356(1-2):65-71.	2015	2.1	
Costello syndrome	Leoni C, Stevenson DA, Martini L, De Sanctis R, Mascolo G, Pantaleoni F, De Santis S, La Torraca I, Persichilli S, Caradonna P, Tartaglia M, Zampino G.	Decreased bone mineral density in Costello syndrome.	Mol Genet Metab. 2014 Jan;111(1):41-5.	2014	3.0	

Partecipazione alle Reti Europee di Riferimento per le Malattie genetiche infantili



Malattie neurologiche e neuromuscolari pediatriche

A cura della Società Italiana di Neuropsichiatria dell'Infanzia e dell'Adolescenza - SINPIA
*(*Presidente: *Antonella Costantino)*

e della Società Italiana di Neurologia Pediatrica - SINP
*(*Presidente*: Raffaele Falsaperla)*

Salvatore De Masi
Renzo Guerrini
Vincenzo Leuzzi
Carlo Minetti
Anna Rosati

Pubblicazioni

MALATTIE NEUROLOGICHE E NEUROMUSCOLARI PEDIATRICHE

Anna Rosati, Salvatore De Masi, Renzo Guerrini
Centro di Eccellenza in Neuroscienze, Dipartimento NEURFARBA – Università degli Studi di Firenze, Azienda Ospedaliero-Universitaria A. Meyer, Firenze

Dal 1 gennaio 2014 ad oggi la produzione scientifica del Centro di Eccellenza in Neuroscienze dell'Azienda Ospedaliero-Universitaria (AOU) Meyer di Firenze include 193 lavori indicizzati su PubMed. La collaborazione del Centro di Eccellenza con centri di ricerca internazionali è documentata da una serie di lavori scientifici e dalla partecipazione a network (European Reference Network on epilepsies) e progetti di ricerca europei ed internazionali (DESIRE, EuroEPINOMICS-RES e PrioMedChild).
In base ai criteri di selezione (malattia neurologica e neuromuscolare rara con e senza codice di esenzione in Italia) e pubblicazione con leadership italiana, 101 articoli sono stati selezionati dagli iniziali 193. In 62/101 lavori il Centro di Eccellenza in Neuroscienze ha rivestito ruolo di leadership.
La maggior produzione scientifica del Centro di Eccellenza in Neuroscienze è rivolta a patologie neurologiche in cui l'epilessia è il sintomo prevalente, ovvero le encefalopatie epilettiche (EE). Si tratta di condizioni rare di interesse quasi esclusivamente pediatrico e comunque ad esordio in età neonatale-infantile, la cui causa può essere malformativa (displasie corticali, polimicrogiria, ecc) o metabolica (malattie mitocondriali, ecc), su base genetica nota o presunta tale, o prettamente genetica, con interessamento di geni coinvolti nel normale funzionamento neuronale. La diagnosi di queste condizioni neurologiche rare è facilitata dall'impiego di un pannello comprendente i geni coinvolti nelle EE e nelle epilessie farmacoresistenti, effettuato presso il Laboratorio di Neurogenetica dell'AOU Meyer, cui fanno riferimento numerosi Centri italiani pediatrici e non.
In base alle caratteristiche elettro-cliniche e all'età d'esordio dell'epilessia, le EE possono essere distinte in sindromi (sindrome di Ohtahara, Encefalopatia mioclonica precoce, Sindrome di West, Sindrome di Dravet, Sindrome di Lennox-Gastuat). In alcuni casi i criteri elettro-clinici non consentono una chiara definizione sindromica dell'EE, la cui causa può inoltre restare sconosciuta. In tali condizioni ancor più difficile appare il tentativo di riconoscimento della condizione quale malattia rara. Diciannove delle 31 pubblicazioni riguardano EE che non sono ad oggi ancora presenti in Allegato 7.
Le malformazioni cerebrali ed in particolare quelle dello sviluppo corticale sono alla base delle EE e dell'epilessia farmacoresistente, quest'ultima ad esordio in qualsiasi fascia d'età pediatrica. Tra i diversi quadri malformativi cerebrali solo alcuni sono presenti nel registro Orphanet e ancora meno in Allegato 7.
Nell'ambito delle patologie metaboliche, la ricerca scientifica verte soprattutto sulla malattia di Fabry, la malattia di Pompe e le patologie mitocondriali.
Vi è infine una discreta attività di ricerca nel campo della malattie vascolari e delle patologie di interesse neurochirurgico e oculistico.

Pubblicazioni

Centro di Eccellenza in Neuroscienze, Dipartimento NEURFARBA – Università degli Studi di Firenze, Azienda Ospedaliero-Universitaria A. Meyer, Firenze

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Spina Bifida	**Giordano F**, Spacca B, Danti A, Taverna M, Losi S, Stagi S, **Genitori L**.	Amenorrhea after Endoscopic Third Ventriculostomy for a Failed Shunt in Spina Bifida: Case Report and Review of the Literature.	Pediatr Neurosurg. 51(1):35-41.	2016	0,24	Non presente in allegato 7
Craniosynostosis	Biamino E, Canale A, Lacilla M, Marinosci A, Dagna F, Genitori L, Peretta P, Silengo M, Albera R, Ferrero GB.	Prevention and management of hearing loss in syndromic craniosynostosis: A case series.	Int J Pediatr Otorhinolaryngol. 85:95-8.	2016	0,90	First and corrisponding authors from University of Torino, Torino, Italy.
cobalamin C defect	**Bacci GM**, Donati MA, Pasquini E, Munier F, Cavicchi C, Morrone A, Sodi A, Murro V, Garcia Segarra N, Defilippi C, Bussolin L, **Caputo R**.	Optical coherence tomography morphology and evolution in cblC disease-related maculopathy in a case series of very young patients.	Acta Ophthalmol.	2017	3,16	
OTX2 gene	Lonero A, Delvecchio M, Primignani P, Caputo R, Bargiacchi S, Penco S, Mauri L, Andreucci E, Faienza MF, Cavallo L.	A novel OTX2 gene frameshift mutation in a child with microphthalmia, ectopic pituitary and growth hormone deficiency.	J Pediatr Endocrinol Metab. 29(5):603-605.	2016	0,91	First and corresponding authors from University of Bari, Bari, Italy.

ROP	Cavallaro G, Filippi L, Bagnoli P, La Marca G, Cristofori G, Raffaeli G, Padrini L, Araimo G, Fumagalli M, Groppo M, Dal Monte M, Osnaghi S, Fiorini P, Mosca F.	The pathophysiology of retinopathy of prematurity: an update of previous and recent knowledge.	Acta Ophthalmol. 92(1):2-20.	2014	3,16	First author from Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico - Università degli Studi di Milano, Milan, Italy. Non presente in allegato 7
Retinoschisis	Murro V, Caputo R, Bacci GM, Sodi A, Mucciolo DP, Bargiacchi S, Giglio SR, Virgili G, Rizzo S.	Case report of an atypical early onset X-linked retinoschisis in monozygotic twins.	BMC Ophthalmol. 17(1):19.	2017	1,59	First and corresponding authors from University of Florence, Florence, Italy.
Severe infantile hemangiomas	**Greco A**, D'Erme AM, Zamma Gallarati B, Caputo R, de Martino M.	A further experience of propranolol for severe infantile hemangiomas of the face: an observational study.	Dermatol Ther. 27(4):198-202.	2014	1,09	Non presente in allegato 7
Uveitis	**Brambilla A**, Caputo R, Cimaz R, **Simonini G**.	Canakinumab for Childhood Sight-threatening Refractory Uveitis: A Case Series.	J Rheumatol. 43(7):1445-1447.	2016	1,66	Non presente in allegato 7
Adenosine Deaminase (ADA) deficiency	Sauer AV, Hernandez RJ, Fumagalli F, Bianchi V, Poliani PL, Dallatomasina C, Riboni E, Politi LS, Tabucchi A, Carlucci F, Casiraghi M, Carriglio N, Cominelli M, Forcellini CA, Barzaghi F, Ferrua F, Minicucci F, Medaglini S, Leocani L, la Marca G, Notarangelo LD, Azzari C, Comi G, Baldoli C, Canale S, Sessa M, D'Adamo P, Aiuti A.	Alterations in the brain adenosine metabolism cause behavioral and neurological impairment in ADA-deficient mice and patients.	Sci Rep. 2017 Jan 11;7:40136.	2017	4,26	First and correspondig authors from IRCCS San Raffaele Hospital, Milan, Italy. Non presente in allegato 7

Fabry disease	Verrecchia E, Zampetti A, Antuzzi D, Ricci R, Ferri L, Morrone A, Feliciani C, Dagna L, Manna R.	The impact of fever/hyperthermia in the diagnosis of Fabry: A retrospective analysis.	Eur J Intern Med. 32:26-30.	2016	0,19	First and correspondig authors from A. Gemelli Policlinic, Catholic University of the Sacred Heart, Rome, Italy.
Fabry disease	Romani I, Borsini W, Nencini P, Morrone A, Ferri L, Frusconi S, Donadio VA, Liguori R, Donati MA, Falconi S, Pracucci G, Inzitari D.	De novo Diagnosis of Fabry Disease among Italian Adults with Acute Ischemic Stroke or Transient Ischemic Attack.	J Stroke Cerebrovasc Dis. 24(11):2588-2595.	2015	1,01	First and correspondig authors from NEUROFARBA Department, University of Florence, Italy.
Adenosine kinase deficiency	Staufner C, Lindner M, Dionisi-Vici C, Freisinger P, Dobbelaere D, Douillard C, Makhseed N, Straub BK, Kahrizi K, Ballhausen D, la Marca G, Kölker S, Haas D, Hoffmann GF, Grünert SC, Blom HJ.	Adenosine kinase deficiency: expanding the clinical spectrum and evaluating therapeutic options.	J Inherit Metab Dis. 39(2):273-283.	2016	3,31	First author from University Hospital Heidelberg, Heidelberg, Germany and corresponding author from University Hospital Freiburg, Germany.
GM1-gangliosidosis	Deodato F, Procopio E, Rampazzo A, Taurisano R, Donati MA, Dionisi-Vici C, Caciotti A, Morrone A, Scarpa M	The treatment of juvenile/adult GM1-gangliosidosis with Miglustat may reverse disease progression.	Metab Brain Dis. 2017 Jun 3.	2017	2,30	First author from Bambino Gesù Children's Hospital, IRCCS, Rome, Italy and corresponding author from University Children's Hospital, Padua, Italy.
GM1-gangliosidosis and Morquio disease type B	Front S, Biela-Banaś A, Burda P, Ballhausen D, Higaki K, Caciotti A, Morrone A,	(5aR)-5a-C-Pentyl-4-epi-isofagomine: A powerful inhibitor of lysosomal β-galactosidase and a remarkable	Eur J Med Chem.126:160-170.	2017	3,18	First and correspondig authors from

	Charollais-Thoenig J, Gallienne E, Demotz S, Martin OR.	chaperone for mutations associated with GM1-gangliosidosis and Morquio disease type B.				Institut de Chimie Organique et Analytique (ICOA), France.
Mitochondrial disease	Mancuso M, Orsucci D, Angelini C, Bertini E, Carelli V, Comi GP, Donati MA, Federico A, Minetti C, Moggio M, Mongini T, Santorelli FM, Servidei S, Tonin P, Toscano A, Bruno C, Bello L, Ienco EC, Cardaioli E, Catteruccia M, Da Pozzo P, Filosto M, Lamperti C, Moroni I, Musumeci O, Pegoraro E, Ronchi D, Sauchelli D, Scarpelli M, Sciacco M, Valentino ML, Vercelli L, Zeviani M, Siciliano G.	Redefining phenotypes associated with mitochondrial DNA single deletion.	J Neurol. 262(5):1301-9.	2015	3,39	First and correspondig authors from University of Pisa, Pisa, Italy.
Mitochondrial disease	Nesti C, Meschini MC, Meunier B, Sacchini M, Doccini S, Romano A, Petrillo S, Pezzini I, Seddiki N, Rubegni A, Piemonte F, Donati MA, Brasseur G, Santorelli FM.	Additive effect of nuclear and mitochondrial mutations in a patient with mitochondrial encephalomyopathy.	Hum Mol Genet. 24(11):3248-3256.	2015	6,35	First and correspondig authors from IRCCS Stella Maris, Pisa, Italy.
Mitochondrial disease	Diodato D, Invernizzi F, Lamantea E, Fagiolari G, Parini R, Menni F, Parenti G, Bollani L, Pasquini E, Donati MA, Cassandrini D, Santorelli FM, Haack TB, Prokisch H, Ghezzi D, Lamperti C, Zeviani M.	Common and Novel TMEM70 Mutations in a Cohort of Italian Patients with Mitochondrial Encephalocardiomyopathy.	JIMD Rep. 15:71-8.	2015	3,36	First and correspondig authors from Fondazione Istituto Neurologico "Carlo Besta", Istituto di Ricovero e Cura a Carattere Scientifico (IRCCS), Milan, Italy.

Pompe Disease	Musumeci O, la Marca G, Spada M, Mondello S, Danesino C, Comi GP, Pegoraro E, Antonini G, Marrosu G, Liguori R, Morandi L, Moggio M, Massa R, Ravaglia S, Di Muzio A, Filosto M, Tonin P, Di Iorio G, Servidei S, Siciliano G, Angelini C, Mongini T, Toscano A; Italian GSD II group.	LOPED study: looking for an early diagnosis in a late-onset Pompe disease high-risk population.	J Neurol Neurosurg Psychiatry. 87(1):5-11.	2016	2,85	First and correspondig authors from Department of Neurosciences, University of Messina, Messina, Italy.
Pompe Disease	Astrea G, Perazza S, Frosini S, Moretti E, Sacchini M, Dati E, Pecini C, Procopio E, Santorelli FM, Donati MA, Battini R.	Infantile-Onset Pompe Disease: The Care Beyond the Cure.	J Neuromuscul Dis. 2(s1):S58-S59.	2015	0,55	First and correspondig authors from IRCCS Fondazione Stella Maris, Calambrone, Italy.
Spastic paraplegia SPG9	Panza E, Escamilla-Honrubia JM, Marco-Marín C, Gougeard N, De Michele G Morra VB, Liguori R, Salviati L, Donati MA, Cusano R, Pippucci T, Ravazzolo R, Németh AH, Smithson S, Davies S, Hurst JA, Bordo D, Rubio V, Seri M.	ALDH18A1 gene mutations cause dominant spastic paraplegia SPG9: loss of function effect and plausibility of a dominant negative mechanism.	Brain. 139(Pt 1):e3.	2016	10,29	First and correspondig authors from University of of Bologna, Italy.
Primary central nervous system vasculitis	**Rosati A**, Cosi A, Basile M, Brambilla A, Guerrini R, Cimaz R, **Simonini G**.	Mycophenolate mofetil as induction and long-term maintaining treatment in childhood: Primary angiitis of the central nervous system.	Joint Bone Spine. 84(3):353-356.	2017	2,95	Non presente in allegato 7

Kaposiform Hemangioendothelioma	**Filippi L**, Tamburini A, Berti E, Perrone A, Defilippi C, Favre C, Calvani M, Della Bona ML, la Marca G, **Donzelli G.**	Successful Propranolol Treatment of a Kaposiform Hemangioendothelioma Apparently Resistant to Propranolol.	Pediatr Blood Cancer. 63(7):1290-1292.	2016	2,63	Non presente in allegato 7
ROP	**Filippi L**, Cavallaro G, Bagnoli P, Dal Monte M, Fiorini P, Berti E, Padrini L, Donzelli G, Araimo G, Cristofori, Fumagalli M, la Marca G, Della Bona ML, Pasqualetti R, Fortunato P, Osnaghi S, Tomasini B, Vanni M, Calvani AM, Milani S, Cortinovis I, Pugi A, Agosti M, Mosca F.	Propranolol 0.1% eye micro-drops in newborns with retinopathy of prematurity: a pilot clinical trial.	Pediatr Res. 81(2):307-314.	2017	2,76	Non presente in allegato 7
Absence Epilepsy	**Guerrini R**, Melani F, Brancati C, Ferrari AR, Brovedani P, Biggeri A, Grisotto L, **Pellacani S**.	Dysgraphia as a Mild Expression of Dystonia in Children with Absence Epilepsy.	PLoS One. 10(7):e0130883.	2015	3,54	Non presente in allegato 7
Autism-epilepsy	Ambrosini E, Sicca F, Brignone MS, D'Adamo MC, Napolitano C, Servettini I, Moro F, Ruan Y, Guglielmi L, Pieroni S, Servillo G, Lanciotti A, Valvo G, Catacuzzeno L, Franciolini F, Molinari P, Marchese M, Grottesi A, Guerrini R, Santorelli FM, Priori S, Pessia M.	Genetically induced dysfunctions of Kir2.1 channels: implications for short QT3 syndrome and autism-epilepsy phenotype.	Hum Mol Genet. 23(18):4875-4886.	2014	6,35	Forst author from Istituto Superiore di Sanità, Rome, Italy and corresponding author from Faculty of Medicine, Rome, Italy. Non presente in allegato 7
Autism-epilepsy	Marchese M, Conti V, Valvo G, Moro F, Muratori F, Tancredi R, Santorelli FM, Guerrini R, Sicca F.	Autism-epilepsy phenotype with macrocephaly suggests PTEN, but not GLIALCAM, genetic screening.	BMC Med Genet. 15:26.	2014	2,38	First and correspondig authors from IRCCS Stella Maris, Pisa, Italy. Non presente in allegato 7

Autism-epilepsy	Chilosi AM, Brovedani P, Ferrari AR, Ziegler AL, Guerrini R, Deonna T.	Language regression associated with autistic regression and electroencephalographic (EEG) abnormalities: a prospective study.	J Child Neurol. 29(6):855-859.	2014	1,43	First author from IRCCS Stella Maris, Pisa, Italy and correspondig author from Unité de Neurologie et de Neuroréhabilitation Pédiatrique CHUV, Lausanne, Switzerland. Non presente in allegato 7
CHRNA2 gene mutations	**Conti V**, Aracri P, Chiti L, Brusco S, Mari F, Marini C, Albanese M, Marchi A, Liguori C, Placidi F, Romigi A, Becchetti A, **Guerrini R**.	Nocturnal frontal lobe epilepsy with paroxysmal arousals due to CHRNA2 loss of function	Neurology. 84(15):1520-1528.	2015	8,17	Non presente in allegato 7
Cortical malformations	**Lenge M**, Barba C, Montanaro D, Aghakhanyan G, Frijia F, **Guerrini R**.	Relationships Between Morphologic and Functional Patterns in the Polymicrogyric Cortex.	Cereb Cortex. 18:1-11.	2017	8,28	
Cortical malformations	Anand S, Cicchi R, Giordano F, Conti V, Buccoliero AM, Guerrini R, Pavone FS.	Multimodal fiber-probe spectroscopy allows detecting epileptogenic focal cortical dysplasia in children.	J Biophotonics.	2017	3,82	First and correspondig authors from University of Florence
Cortical malformations	**Parrini E**, Conti V, Dobyns WB, **Guerrini R.**	Genetic Basis of Brain Malformations	Mol Syndromol. 7(4):220-233.	2016		
Cortical malformations	**Bartolini E**, Falchi M, Zellini F, Parrini E, Grisotto L, CosottiniM, Posar A, Parmeggiani A, Ambrosetto G, Ferrari AR, Santucci M, Salas-Puig J, Barba C, **Guerrini R**.	The syndrome of polymicrogyria, thalamic hypoplasia, and epilepsy with CSWS.	Neurology. 86(13):1250-1259.	2016	8,17	

Cortical malformations	Mirzaa GM, Conti V, Timms AE, Smyser CD, Ahmed S, Carter M, Barnett S, Hufnagel RB, Goldstein A, Narumi-Kishimoto Y, Olds C, Collins S, Johnston K, Deleuze JF, Nitschké P, Friend K, Harris C, Goetsch A, Martin B, Boyle EA, Parrini E, Mei D, Tattini L, Slavotinek A, Blair E, Barnett C, Shendure J, Chelly J, Dobyns WB, **Guerrini R**.	Characterisation of mutations of the phosphoinositide-3-kinase regulatory subunit, PIK3R2, in perisylvian polymicrogyria: a next-generation sequencing study.	Lancet Neurol. 14(12):1182-1195.	2015	23,47	
Cortical malformations	Barkovich AJ, Dobyns WB, **Guerrini R**.	Malformations of cortical development and epilepsy.	Cold Spring Harb Perspect Med. 5(5):a022392.	2015	9,47	
Cortical malformations	**Buccoliero AM**, Barba C, Giordano F, Baroni G, Genitori L, Guerrini R, Taddei GL.	Expression of glutamine synthetase in balloon cells: a basis of their antiepileptic role?	Clin Neuropathol. 34(2):83-88.	2015	1,07	
Cortical malformations	**De Ciantis A**, Barkovich AJ, Cosottini M, Barba C, Montanaro D, Costagli M, Tosetti M, Biagi L, Dobyns WB, **Guerrini R**.	Ultra-high-field MR imaging in polymicrogyria and epilepsy.	AJNR Am J Neuroradiol. 36(2):309-316.	2015	3,12	
Cortical malformations	Capra V, Biancheri R, Morana G, Striano P, Novara F, Ferrero GB, Boeri L, Celle ME, Mancardi MM, Zuffardi O, Parrini E, **Guerrini R**.	Periventricular nodular heterotopia in Smith-Magenis syndrome.	Am J Med Genet A. 164A(12):3142-147.	2014	2,08	
Cortical malformations	**Conti V**, Pantaleo M, Barba C, Baroni G, Mei D, Buccoliero AM, Giglio S, Giordano F, Baek ST, Gleeson JG, **Guerrini R**.	Focal dysplasia of the cerebral cortex and infantile spasms associated with somatic 1q21.1-q44 duplication including the AKT3 gene.	Clin Genet. 88(3):241-247.	2015	3,89	

Cortical malformations	**Guerrini R**, Dobyns WB.	Malformations of cortical development: clinical features and genetic causes.	Lancet Neurol. 13(7):710-726.	2014	23,47	
Cortical malformations	**Barba C**, Parrini E, Coras R, Galuppi A, Craiu D, Kluger G, Parmeggiani A, Pieper T, Schmitt-Mechelke T, Striano P, Giordano F, Blumcke I, **Guerrini R**.	Co-occurring malformations of cortical development and SCN1A gene mutations.	Epilepsia. 55(7):1009-1019.	2014	4,71	
Dravet syndrome	**Cetica V**, Chiari S, Mei D, Parrini E, Grisotto L, Marini C, Pucatti D, Ferrari A, Sicca F, Specchio N, Trivisano M, Battaglia D, Contaldo I, Zamponi N, Petrelli C, Granata T, Ragona F, Avanzini G, **Guerrini R**.	Clinical and genetic factors predicting Dravet syndrome in infants with SCN1A mutations.	Neurology. 88(11):1037-1044.	2017	8,17	
SAWNTAMARIADravet syndrome	**Guerrini R**, Striano P.	Dravet syndrome: Not just epilepsy.	Neurology. 87(3):245-246.	2016	8,17	
Dravet syndrome	Doccini S, Meschini MC, Mei D, Guerrini R, Sicca F, Santorelli FM.	Mitochondrial respiratory chain defects in skin fibroblasts from patients with Dravet syndrome.	Neurol Sci. 36(11):2151-2155.	2015	1,48	First and correspondig authors from IRCCS Stella Maris, Pisa, Italy.
Drug-resistant epilepsy	**Parrini E**, Marini C, Mei D, Galuppi A, Cellini E, Pucatti D, Chiti L, Rutigliano D, Bianchini C, Virdò S, De Vita D, Bigoni S, Barba C, Mari F, Montomoli M, Pisano T, Rosati A; ClinicalStudy Group, **Guerrini R**.	Diagnostic Targeted Resequencing in 349 Patients with Drug-Resistant Pediatric Epilepsies Identifies Causative Mutations in 30 Different Genes.	Hum Mutat. 38(2):216-225.	2017	5,09	Non presente in allegato 7

Drug-resistant epilepsy	De Liso P, Vigevano F, Specchio N, De Palma L, Bonanni P, Osanni E, Coppola G, Parisi P, Grosso S, Verrotti A, Spalice A, Nicita F, Zamponi N, Siliquini S, Giordano L, Martelli P, Guerrini R, Rosati A, Ilvento L, Belcastro V, Striano P, Vari MS, Capovilla G, Beccaria F, Bruni O, Luchetti A, Gobbi G, Russo A, Pruna D, Tozzi AE, Cusmai R.	Effectiveness and tolerability of perampanel in children and adolescents with refractory epilepsies-An Italian observational multicenter study.	Epilepsy Res. 127:93-100.	2016	2,75	First and correspondig authors from Bambino Gesù Children's Hospital, IRCCS of Rome. Non presente in allegato 7
Drug-resistant epilepsy	Franco V, Canevini MP, Capovilla G, De Sarro G, Galimberti CA, Gatti G, Guerrini R, La Neve A, Rosati E, Specchio LM, Striano S, Tinuper P, Perucca E.	Off-label prescribing of antiepileptic drugs in pharmacoresistant epilepsy: a cross-sectional drug utilization study of tertiary care centers in Italy.	CNS Drugs. 28(10):939-949.	2014	4,91	First and correspondig authors from University of Pavia, Pavia, Italy. Non presente in allegato 7
Epileptic encephalopathies	**Barba C**, Darra F, Cusmai R, Procopio E, Dionisi Vici C, Keldermans L, Vuillaumier-Barrot S, Lefeber DJ, **Guerrini R**; CDG Group.	Congenital disorders of glycosylation presenting as epileptic encephalopathy with migrating partial seizures in infancy.	Dev Med Child Neurol. 58(10):1085-1091.	2016	3,61	Non presente in allegato 7
Epileptic encephalopathies	**Mei D**, Darra F, Barba C, Marini C, Fontana E, Chiti L, Parrini E, Dalla Bernardina B, **Guerrini R**.	Optimizing the molecular diagnosis of CDKL5 gene-related epileptic encephalopathy in boys.	Epilepsia. 55(11):1748-1753.	2014	4,71	Non presente in allegato 7
FLNA gene mutations	**Parrini E**, Mei D, Pisanti MA, Catarzi S, Pucatti D, Bianchini C, Mascalchi M, Bertini E, Morrone A, Cavaliere ML, **Guerrini R**.	Familial periventricular nodular heterotopia, epilepsy and Melnick-Needles Syndrome caused by a single FLNA mutation with combined gain-of-function and loss-of-function effects.	J Med Genet. 52(6):405-412.	2015	5,65	Non presente in allegato 7

FOXG1 gene mutations	**Cellini E**, Vignoli A, Pisano T, Falchi M, Molinaro A, Accorsi P, Bontacchio A, Pinelli L, Giordano L, **Guerrini R**; FOXG1 Syndrome Study Group.	The hyperkinetic movement disorder of FOXG1-related epileptic-dyskinetic encephalopathy.	Dev Med Child Neurol. 58(1):93-97.	2016	3,61	Non presente in allegato 7
GNAO1 encephalopathy	Danti FR, Galosi S, Romani M, Montomoli M, Carss KJ, Raymond FL, Parrini E, Bianchini C, McShane T, Dale RC, Mohammad SS, Shah U, Mahant N, Ng J, McTague A, Samanta R, Vadlamani G, Valente EM, Leuzzi V, Kurian MA, **Guerrini R**.	GNAO1 encephalopathy: Broadening the phenotype and evaluating treatment and outcome.	Neurol Genet. 3(2):e143	2017		Non presente in allegato 7
Histiocytosis	**Sieni E**, Barba C, Mortilla M, Savelli S, Grisotto L, Di Giacomo G, Romano K, Fonda C, Biggeri A, Guerrini R, Aricò M**.**	Early Diagnosis and Monitoring of Neurodegenerative Langerhans Cell Histiocytosis.	PLoS One. 10(7):e0131635.	2015	3,54	
Hypermotor epilepsy	Tinuper P, Bisulli F, Cross JH, Hesdorffer D, Kahane P, Nobili L, Provini F, Scheffer IE, Tassi L, Vignatelli L, Bassetti C, Cirignotta F, Derry C, Gambardella A, Guerrini R, Halasz P, Licchetta L, Mahowald M, Manni R, Marini C, Mostacci B, Naldi I, Parrino L, Picard F, Pugliatti M, Ryvlin P, Vigevano F,Zucconi M, Berkovic S, Ottman R.	Definition and diagnostic criteria of sleep-related hypermotor epilepsy.	Neurology. 86(19):1834-1842.	2016	8,17	First author from University of Bologna, Italy and correspondig author from Columbia University, New York. Non presente in allegato 7

Hypoxic ischemic encephalopathy	**Filippi L**, Fiorini P, Catarzi S, Berti E, Padrini L, Landucci E, Donzelli G, Bartalena L, Fiorentini E, Boldrini A, Giampietri M, Scaramuzzo RT, la Marca G, Della Bona ML, Fiori S, TinelliF, Bancale A, Guzzetta A, Cioni G, Pisano T, Falchi M, **Guerrini R**.	Safety and efficacy of topiramate in neonates with hypoxic ischemic encephalopathy treated with hypothermia (NeoNATI): a feasibility study.	J Matern Fetal Neonatal Med. 28:1-8.	2017	1,67	Non presente in allegato 7
Juvenile myoclonic epilepsy	Mumoli L, Tarantino P, Michelucci R, Bianchi A, Labate A, Franceschetti S, Marini C, Striano P, Gagliardi M, Ferlazzo E, Sofia V, Pennese L, Annesi G, Aguglia U, Guerrini R, Zara F, Gambardella A; Genetic Commission, Italian League Against Epilepsy.	No evidence of a role for cystatin B gene in juvenile myoclonic epilepsy.	Epilepsia. 56(4):e40-43.	2015	4,71	First and correspondig authors from University Magna Graecia Catanzaro, Catanzaro, Italy. Non presente in allegato 7
KCNQ2 gene mutations	**Pisano T**, Numis AL, Heavin SB, Weckhuysen S, Angriman M, Suls A, Podesta B, Thibert RL, Shapiro KA, Guerrini R, Scheffer IE, Marini C, Cilio MR.	Early and effective treatment of KCNQ2 encephalopathy.	Epilepsia. 56(5):685-691.	2015	4,71	Non presente in allegato 7
Mutations in OPA1	Rubegni A, Pisano T, Bacci G, Tessa A, Battini R, Procopio E, Giglio S, Pasquariello R, Santorelli FM, Guerrini R, Nesti C.	Leigh-like neuroimaging features associated with new biallelic mutations in OPA1	Eur J Paediatr Neurol. 21(4):671-677	2017	2,12	First and correspondig authors from IRCCS Stella Maris, Pisa. Non presente in allegato 7

Ring chromosome 20 syndrome	Vignoli A, Bisulli F, Darra F, Mastrangelo M, Barba C, Giordano L, Turner K, Zambrelli E, Chiesa V, Bova S, Fiocchi I, Peron A, Naldi I, Milito G, Licchetta L, Tinuper P, Guerrini R, Dalla Bernardina B, Canevini MP.	Epilepsy in ring chromosome 20 syndrome.	Epilepsy Res. 128:83-93.	2016	2,75	First and correspondig authors from San Paolo Hospital of Milano.
SCN1A gene mutations	Passamonti C, Petrelli C, Mei D, Foschi N, Guerrini R, Provinciali L, Zamponi N.	A novel inherited SCN1A mutation associated with different neuropsychological phenotypes: is there a common core deficit?	Epilepsy Behav. 43:89-92.	2015	0,92	First and correspondig authors from Department of Neuropsychiatry, Ospedali Riuniti, Ancona, Italy.
SLC35A3 encephalopathy	**Marini C**, Hardies K, Pisano T, May P, Weckhuysen S, Cellini E, Suls A, Mei D, Balling R, Jonghe PD, Helbig I, Garozzo D; EuroEPINOMICS consortium AR working group, **Guerrini R**.	Recessive mutations in SLC35A3 cause early onset epileptic encephalopathy with skeletal defects.	Am J Med Genet. 173(4):1119-1123	2017	2,08	Non presente in allegato 7
Spasms	**Barba C**, Mai R, Grisotto L, Gozzo F, Pellacani S, Tassi L, Francione S, Giordano F, Cardinale F, **Guerrini R**.	Unilobar surgery for symptomatic epileptic spasms.	Ann Clin Transl Neurol. 4(1):36-45.	2016		
Spasms	Ronzano N, Valvo G, Ferrari AR, Guerrini R, Sicca F.	Late-onset epileptic spasms: clinical evidence and outcome in 34 patients.	J Child Neurol. 2015 Feb;30(2):153-9	2015	1,43	First and correspondig authors from IRCCS Stella Maris, Pisa, Italy.

Status epilepticus	**Rosati A**, Ilvento L, L'Erario M, De Masi S, Biggeri A, Fabbro G, Bianchi R, Stoppa F, Fusco L, Pulitanò S, Battaglia D, Pettenazzo A, Sartori S, Biban P, Fontana E, Cesaroni E, MoraD, Costa P, Meleleo R, Vittorini R, Conio A, Wolfler A, Mastrangelo M, Mondardini MC, Franzoni E, McGreevy KS, Di Simone L, Pugi A, Mirabile L, Vigevano F, **Guerrini R**.	Efficacy of ketamine in refractory convulsive status epilepticus in children: a protocol for a sequential design, multicentre, randomised, controlled, open-label, non-profit trial (KETASER01)	BMJ Open. 6(6):e011565.	2016	2,56	Non presente in allegato 7
Status epilepticus	**Ilvento L**, Rosati A, Marini C, L'Erario M, Mirabile L, **Guerrini R.**	Ketamine in refractory convulsive status epilepticus in children avoids endotracheal intubation	Epilepsy Behav. 49:343-346.	2015	0,92	Non presente in allegato 7
Symptomatic focal epilepsies	Vecchi M, Barba C, De Carlo D, Stivala M, Guerrini R, Albamonte E, Ranalli D, Battaglia D, Lunardi G, Boniver C, Piccolo B, Pisani F, Cantalupo G, Nieddu G, Casellato S, Cappanera S, Cesaroni E, Zamponi N, Serino D, Fusco L, Iodice A, Palestra F, Giordano L, Freri E, De Giorgi I, Ragona F, Granata T, Fiocchi I, Bova SM, Mastrangelo M, Verrotti A, Matricardi S, Fontana E, Caputo D, Darra F, Dalla Bernardina B, Beccaria F, Capovilla G, Baglietto MP, Gagliardi A, Vignoli A, Canevini MP, Perissinotto E, Francione S.	Symptomatic and presumed symptomatic focal epilepsies in childhood: An observational, prospective multicentre study.	Epilepsia. 57(11):1808-1816.	2016	4,71	First author from University of Padua and corresponding author from Niguarda Hospital of Milan.

TITF1 gene mutations	**Rosati A**, Berti B, Melani F, Cellini E, Procopio E, **Guerrini R**.	Recurrent drop attacks in early childhood as presenting symptom of benign hereditary chorea caused by TITF1 gene mutations.	Dev Med Child Neurol. 57(8):777-779.	2015	3,61	Non presente in allegato 7
Duchenne Muscular Dystrophy	Mazzone ES, Coratti G, Sormani MP, Messina S, Pane M, D'Amico A, Colia G, Fanelli L, Berardinelli A, Gardani A, Lanzillotta V, D'Ambrosio P, Petillo R, Cavallaro F, Frosini S, Bello L, Bonfiglio S, De Sanctis R, Rolle E, Forcina N, Magri F, Vita G, Palermo C, Donati MA, Procopio E, Arnoldi MT, Baranello G, Mongini T, Pini A, Battini R, Pegoraro E, Torrente Y, Previtali SC, Bruno C, Politano L, Comi GP, D'Angelo MG, Bertini E, Mercuri E.	Timed Rise from Floor as a Predictor of Disease Progression in Duchenne Muscular Dystrophy: An Observational Study.	PLoS One. 11(3):e0151445.	2016	3,54	First and corresponding author from Catholic University, Rome, Italy.
Duchenne Muscular Dystrophy	Pane M, Mazzone ES, Sivo S, Sormani MP, Messina S, D'Amico A, Carlesi A, Vita G, Fanelli L, Berardinelli A, Torrente Y, Lanzillotta V, Viggiano E, D'Ambrosio P, Cavallaro F, Frosini S, Barp A, Bonfiglio S, Scalise R, De Sanctis R, Rolle E, Graziano A, Magri F, Palermo C, Rossi F, Donati MA, Sacchini M, Arnoldi MT, Baranello G, Mongini T, Pini A, Battini R, Pegoraro E, Previtali S, Bruno C, Politano L, Comi GP, Bertini E, Mercuri E.	Correction: Long Term Natural History Data in Ambulant Boys with Duchenne Muscular Dystrophy: 36-Month Changes.	PLoS One. 10(12):e0144079.	2015	3,54	First and corresponding author from Catholic University, Rome, Italy.

Duchenne Muscular Dystrophy	Pane M, Mazzone ES, Sormani MP, Messina S, Vita GL, Fanelli L, Berardinelli A, Torrente Y, D'Amico A, Lanzillotta V, Viggiano E, D'Ambrosio P, Cavallaro F, Frosini S, Bello L, Bonfiglio S, Scalise R, De Sanctis R, Rolle E, Bianco F, Van der Haawue M, Magri F, Palermo C, Rossi F, Donati MA, Alfonsi C, Sacchini M, Arnoldi MT, Baranello G, Mongini T, Pini A, Battini R, Pegoraro E, Previtali SC, Napolitano S, Bruno C, Politano L, Comi GP, Bertini E, Morandi L, Gualandi F, Ferlini A, Goemans N, Mercuri E.	6 Minute walk test in Duchenne MD patients with different mutations: 12 month	PLoS One.9(1):e83400.	2014	3,54	First and corresponding author from Catholic University, Rome, Italy.
Myelitis	Bianchi A, Bartolini E, Melani F, Guerrini R, **Mascalchi M**.	Isolated recurrent myelitis in a 7-year-old child with serum aquaporin-4 IgG antibodies.	J Neurol. 264(1):179-181.	2017	3,41	Non presente in allegato 7
PLA2G6-Associated Infantile Neuronal Axonal Dystrophy	**Mascalchi M**, Mari F, Berti B, Bartolini E, Lenge M, Bianchi A, Antonucci L, Santorelli FM, Garavaglia B, **Guerrini R**.	Fast Progression of Cerebellar Atrophy in PLA2G6-Associated Infantile Neuronal Axonal Dystrophy.	Cerebellum. 16(3):742-745.	2017	2,43	
Brain tumor	**Sardi I**, Fantappiè O, la Marca G, Giovannini MG, Iorio AL, da Ros M, Malvagia S, Cardellicchio S, Giunti L, de Martino M, Mazzanti R.	Delivery of doxorubicin across the blood-brain barrier by ondansetron pretreatment: a study in vitro and in vivo.	Cancer Lett. 53(2):242-247.	2014	6,80	Non presente in allegato 7
Brain tumor	**Cardellicchio S**, Bacci G, Farina S, Genitori L, Massimino M, de Martino M, Caputo R, **Sardi I**.	Low-dose cisplatin-etoposide regimen for patients with optic pathway glioma: a report of four cases and literature review.	Neuropediatrics.45(1):42-49.	2014	1,57	Non presente in allegato 7

Brain tumor	**Giunti L**, Buccoliero AM, Pantaleo M, Lucchesi M, Provenzano A, Palazzo V, Guarducci S, Guidi M, Genitori L, Zuffardi O, Sardi I, **Giglio S**.	Molecular characterization of paediatric glioneuronal tumours with neuropil-like islands: a genome-wide copy number analysis.	Am J Cancer Res. 6(12):2910-2918.	2016	3,26	Non presente in allegato 7
Brain tumor	Massimino M, Miceli R, Giangaspero F, Boschetti L, Modena P, Antonelli M, Ferroli P, Bertin D, Pecori E, Valentini L, Biassoni V, Garrè ML, Schiavello E, Sardi I, Cama A, Viscardi E, Scarzello G, Scoccianti S, Mascarin M, Quaglietta L, Cinalli G, Diletto B, Genitori L, Peretta P, Mussano A, Buccoliero A, Calareso G, Barra S, Mastronuzzi A, Giussani C, Marras CE, Balter R, Bertolini P, Giombelli E, La Spina M, Buttarelli FR, Pollo B, Gandola L.	Final results of the second prospective AIEOP protocol for pediatric intracranial ependymoma.	Neuro Oncol. 18(10):1451-60.	2016	2,98	First and corresponding author from Fondazione IRCCS Istituto Nazionale dei Tumori, Milan, Italy. Non presente in allegato 7
Brain tumor	Iorio AL, Ros Md, Fantappiè O, Lucchesi M, Facchini L, Stival A, Becciani S, Guidi M, Favre C, Martino Md, Genitori L, **Sardi I**.	Blood-Brain Barrier and Breast Cancer Resistance Protein: A Limit to the Therapy of CNS Tumors and Neurodegenerative Diseases.	Anticancer Agents Med Chem. 16(7):810-815.	2016	1,61	Non presente in allegato 7
Brain tumor	**Sardi I**, Lucchesi M, Becciani S, Facchini L, Guidi M, Buccoliero AM, Moriondo M, Baroni G, Stival A, Farina S, Genitori L, **de Martino M**.	Absence of human cytomegalovirus infection in childhood brain tumors.	Am J Cancer Res. 5(8):2476-83.	2015	3,26	Non presente in allegato 7

Brain tumor	**Stival A**, Lucchesi M, Farina S, Buccoliero AM, Castiglione F, Genitori L, de Martino M, **Sardi I**.	An infant with hyperalertness, hyperkinesis, and failure to thrive: a rare diencephalic syndrome due to hypothalamic anaplastic astrocytoma.	BMC Cancer. 15:616.	2015	3,26	Non presente in allegato 7
Brain tumor	**Giunti L**, da Ros M, Vinci S, Gelmini S, Iorio AL, Buccoliero AM, Cardellicchio S, Castiglione F, Genitori L, de Martino M, Giglio S, Genuardi M, **Sardi I**.	Anti-miR21 oligonucleotide enhances chemosensitivity of T98G cell line to doxorubicin by inducing apoptosis.	Am J Cancer Res. 5(1):231-42.	2014	3,26	Non presente in allegato 7
Brain tumor	**Giunti L,** Pantaleo M, Sardi I, Provenzano A, Magi A, Cardellicchio S, Castiglione F, Tattini L, Novara F, Buccoliero AM, de Martino M, Genitori L, Zuffardi O, **Giglio S**.	Genome-wide copy number analysis in pediatric glioblastoma multiforme.	Am J Cancer Res. 4(3):293-303.	2014	3,26	Non presente in allegato 7
Brain tumor	**Cardellicchio S**, Farina S, Buccoliero AM, Agresti B, Genitori L, de Martino M, Fangusaro J, **Sardi I**.	Successful treatment of metastatic βHCG-secreting germ cell tumor occurring 3 years after total resection of a pineal mature teratoma.	Eur J Pediatr. 173(8):1011-5.	2014	1,85	Non presente in allegato 7
Spinal tumor	**Spacca** B, Giordano F, Donati P, **Genitori L**.	Spinal tumors in children: long-term retrospective evaluation of a series of 134 cases treated in a single unit of pediatric neurosurgery.	Spine J. 15(9):1949-1955.	2015	0,41	Non presente in allegato 7

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Nome MR	**Autori**	**Titolo del lavoro**	**Rivista, volume, pagine**	**Anno**	**I. F.**	**Note**
Ataxia	**Barresi S**, Niceta M, Alfieri P, Brankovich V, Piccini G, Bruselles A, Barone MR, Cusmai R, Tartaglia M, Bertini E, Zanni G	Mutations in the IRBIT domain of ITPR1 are a frequent cause of autosomal dominant nonprogressive congenital ataxia	Clin Genet. 91(1):86-91.	2017	3,89	
Ataxia	Calì T, Frizzarin M, Luoni L, Zonta F, Pantano S, Cruz C, Bonza MC, Bertipaglia I, Ruzzene M, De Michelis MI, Damiano N, Marin O, Zanni G, Zanotti G, Brini M, Lopreiato R, Carafoli E	The ataxia related G1107D mutation of the plasma membrane Ca2+ ATPase isoform 3 affects its interplay with calmodulin and the autoinhibition process	Biochim Biophys Acta. 1863(1):165-173.	2017	5,16	First and correspondig authors from University of Padua
Ataxia	Calì T, Lopreiato R, Shimony J, Vineyard M, Frizzarin M, Zanni G, Zanotti G, Brini M, Shinawi M, Carafoli E	A novel mutation in isoform 3 of the plasma membrane Ca²⁺ pump impairs cellular Ca²⁺ homeostasis in a patient with cerebellar ataxia and laminin subunit 1Î± mutations	J Biol Chem. 290(26):16132-41.	2015	4,57	First and correspondig authors from University of Padua
Ataxia	**Carletti B**, Piermarini E, Tozzi G, Travaglini L, Torraco A, Pastore A, Sparaco M, Petrillo S, Carrozzo R, Bertini ES, Piemonte F	Frataxin silencing inactivates mitochondrial complex I in NSC34 motoneuronal cells and alters glutathione homeostasis	Int J Mol Sci. 15(4):5789-806.	2014	2,34	

Ataxia	Germanotta M, Vasco G, Petrarca M, Rossi S, Carniel S, Bertini E, Cappa P, **Castelli E**	Robotic and clinical evaluation of upper limb motor performance in patients with Friedreich's Ataxia: An observational study	J Neuroeng Rehabil. 12:41.	2015	2,74	
Ataxia	Piermarini E, Cartelli D, Pastore A, Tozzi G, Compagnucci C, Giorda E, D'Amico J, Petrini S, Bertini E, Cappelletti G, **Piemonte F**	Frataxin silencing alters microtubule stability in motor neurons: implications for Friedreich's ataxia	Hum Mol Genet. 25(19):4288-4301.	2016	5,99	
Ataxia	**Travaglini L**, Nardella M, Bellacchio E, D'Amico A, Capuano A, Frusciante R, Di Capua M, Cusmai R, Barresi S, Morlino S, Fernández-Fernández JM, Trivisano M, Specchio N, Valeriani M, Vigevano F, Bertini E, Zanni G	Missense mutations of CACNA1A are a frequent cause of autosomal dominant nonprogressive congenital ataxia	Eur J Paediatr Neurol. 21(3):450-456.	2017	1,92	
Ataxia	**Vasco G**, Gazzellini S, Petrarca M, Lispi ML, Pisano A, Zazza M, Della Bella G, Castelli E, Bertini E	Functional and Gait Assessment in Children and Adolescents Affected by Friedreich's Ataxia: A One-Year Longitudinal Study	PLoS One. 11(9):e0162463.	2016	3,06	
Behr syndrome	Carelli V, Sabatelli M, Carrozzo R, Rizza T, Tonon C, Lodi R, **Bertini ES**	Behr syndrome' with OPA1 compound heterozygote mutations	Brain. 138:e321	2015	10,23	

Brain abnormalities	Micalizzi A, Poretti A, Romani M, Ginevrino M, Mazza T, Aiello C, Zanni G, Baumgartner B, Borgatti R, Brockmann K, Camacho A, Cantalupo G, Haeusler M, Hikel C, Klein A, Mandrile G, Mercuri E, Rating D, Romaniello R, Santorelli FM, Schimmel M, Spaccini L, Teber S, von Moers A, Wente S, Ziegler A, Zonta A, Bertini E, Boltshauser E, Valente EM	Clinical, neuroradiological and molecular characterization of cerebellar dysplasia with cysts (Poretti-Boltshauser syndrome)	Eur J Hum Genet. 24(9):1262-7.	2016	4,58	First and corresponding authors from IRCCS Casa Sollievo della Sofferenza Hospital, San Giovanni Rotondo, see MALFORMATIONS AND ID
Brain abnormalities	Parrini E, Mei D, Pisanti MA, Catarzi S, Pucatti D, Bianchini C, Mascalchi M, Bertini E, Morrone A, Cavaliere ML, Guerrini R	Familial periventricular nodular heterotopia, epilepsy and Melnick-Needles Syndrome caused by a single FLNA mutation with combined gain-of-function and loss-of-function effects	J Med Genet. 52(6):405-12.	2015	6,34	First and correspondig authors from Meyer Children's Hospital, Florence, see NEUROLOGICAL
Brain abnormalities	Sarubbo S, De Benedictis A, Merler S, Mandonnet E, Balbi S, Granieri E, Duffau H	Towards a functional atlas of human white matter	Hum Brain Mapp. 36(8):3117-36.	2015	5,97	First and correspondig authors from other institutions
Dystonia	**Marras CE**, Rizzi M, Cantonetti L, Rebessi E, De Benedictis A, Portaluri F, Randi F, Savioli A, Castelli E, Vigevano F	Pallidotomy for medically refractory status dystonicus in childhood	Dev Med Child Neurol. 56(7):649-56.	2014	3,29	

Earlyonset schizophrenia	**Armando M**, Pontillo M, Vicari S	Psychosocial interventions for very early and earlyonset schizophrenia: A review of treatment efficacy	Curr Opin Psychiatry. 28(4):312-23.	2015	3,94	
Gilles de la Tourette syndrome	Prontera P, Napolioni V, Ottaviani V, Rogaia D, Fusco C, Augello B, Serino D, Parisi V, Bernardini L, Merla G, Cavanna A, Donti E	DPP6 gene disruption in a family with Gilles de la Tourette syndrome	Neurogenetics. 15(4):237-42.	2014	2,66	First and corresponding authors from University of Perugia
Menkes disease	Verrotti A., Cusmai R, Darra F, Martelli P, Accorsi P, Bergamo S, Bevivino E, Coppola G, Freri E, Grosso S, Matricardi S, Parisi P, Sartori S, Spalice A, Specchio N, Carelli A, Zini D, Bernardina B, Giordano L	Epilepsy in Menkes disease: An electroclinical long-term study of 28 patients	Epilepsy Res. 108(9):1597-603.	2014	2,19	First and corresponding authors from University of Perugia, see METABOLIC
Movement disorder	Nicita F, Travaglini L, Sabatini S, Garavaglia B, Panteghini C, Valeriani M, Bertini E, Nardocci N, Vigevano F, Capuano A	Childhood-onset ATP1A3-related conditions: Report of two new cases of phenotypic spectrum	Parkinsonism Relat Disord. 30:81-2.	2016	3,79	First and correspondig authors from Sapienza University of Rome
Neurodegenerative Diseases	De Tommaso M, Arendt-Nielsen L, Defrin R, Kunz M, Pickering G, Valeriani M	Pain Assessment in Neurodegenerative Diseases	Behav Neurol. 2016:2949358.	2016	1,63	First and correspondig authors from University of Bari
Neurodegenerative Diseases	De Tommaso M, Arendt-Nielsen L, Defrin R, Kunz M, Pickering G, Valeriani M	Pain in Neurodegenerative Disease: Current Knowledge and Future Perspectives	Behav Neurol. 2016:7576292.	2016	1,63	First and correspondig authors from University of Turin

Neurodegenerative Diseases	Di Giacopo R, Cianetti L, Caputo V, La Torraca I, Piemonte F, Ciolfi A, Petrucci S, Carta C, Mariotti P, Leuzzi V, Valente EM, D'Amico A, Bentivoglio A, Bertini E, Tartaglia M, Zampino G	Protracted late infantile ceroid lipofuscinosis due to TPP1 mutations: Clinical, molecular and biochemical characterization in three sibs	J Neurol Sci. 356(1-2):65-71.	2015	2,47	First and correspondig authors from University of Trento, see METABOLIC
Neurodegenerative Diseases	Lanciotti A, Brignone MS, Visentin S, De Nuccio C, Catacuzzeno L, Mallozzi C, Petrini S, Caramia M, Veroni C, Minnone G, Bernardo A, Franciolini F, Pessia M, Bertini E, Petrucci TC, Ambrosini E	Megalencephalic leukoencephalopathy with subcortical cysts protein-1 regulates epidermal growth factor receptor signaling in astrocytes	Hum Mol Genet. 25(8):1543-58.	2016	5,99	First and correspondig authors from ISS, Rome
Neurodegenerative Diseases	Pastore A, Petrillo S, Piermarini E, Piemonte F	Systemic Redox Biomarkers in Neurodegenerative Diseases	Curr Drug Metab. 16(1):46-70.	2015	2,98	
Neurodegenerative Diseases	Petroni M, Sardina F, Heil C, Sahún-Roncero M, Colicchia V, Veschi V, Albini S, Fruci D, Ricci B, Soriani A, Di Marcotullio L, Screpanti I, Gulino A, Giannini G	The MRN complex is transcriptionally regulated by MYCN during neural cell proliferation to control replication stress	Cell Death Differ. 23(2):197-206.	2016	8,18	First and correspondig authors from Sapienza University of Rome
Neurodegenerative Diseases	Pezzini F, Bettinetti L, Di Leva F, Bianchi M, Zoratti E, Carrozzo R, Santorelli FM, Delledonne M, Lalowski M, Simonati A	Transcriptomic Profiling Discloses Molecular and Cellular Events Related to Neuronal Differentiation in SH-SY5Y Neuroblastoma Cells	Cell Mol Neurobiol.37(4):665-682.	2017	2,33	First and correspondig authors from University of Verona

Neurodegenerative Diseases	**Sferra A,** Baillat G, Rizza T, Barresi S, Flex E, Tasca G, D'Amico A, Bellacchio E, Ciolfi A, Caputo V, Cecchetti S, Torella A, Zanni G, Diodato D, Piermarini E, Niceta M, Coppola A, Tedeschi E, Martinelli D, Dionisi-Vici C, Nigro V, Dallapiccola B, Compagnucci C, Tartaglia M, Haase G, Bertini E	TBCE Mutations Cause Early-Onset Progressive Encephalopathy with Distal Spinal Muscular Atrophy	Am J Hum Genet. 99(4):974-983.	2016	10,79	
Neurodegenerative Diseases	Sieni E, Barba C, Mortilla M, Savelli S, Grisotto L, Di Giacomo G, Romano K, Fonda C, Biggeri A, Guerrini R, Aricò M	Early Diagnosis and Monitoring of Neurodegenerative Langerhans Cell Histiocytosis	PLoS One. 10(7):e0131635.	2015	3,23	First and correspondig authors from Meyer Children's Hospital, Florence
Neurodegenerative Diseases	Tonduti D, Aiello C, Renaldo F, Dorboz I, Saaman S, Rodriguez D, Fettah H, Elmaleh M, Biancheri R, Barresi S, Boccone L, Orcesi S, Pichiecchio A, Zangaglia R, Maurey H, Rossi A, Boespflug-Tanguy O, Bertini E	TUBB4A-related hypomyelinating leukodystrophy: New insights from a series of 12 patients	Eur J Paediatr Neurol. 20(2):323-30.	2016	2,30	First and correspondig authors from Besta Foundation IRCCS, Milan

Neurodegenerative Diseases	**Torraco A**, Ardissone A, Invernizzi F, Rizza T, Fiermonte G, Niceta M, Zanetti N, Martinelli D, Vozza A, Verrigni D, Di Nottia M, Lamantea E, Diodato D, Tartaglia M, Dionisi-Vici C, Moroni I, Farina L, Bertini E, Ghezzi D, Carrozzo R	Novel mutations in IBA57 are associated with leukodystrophy and variable clinical phenotypes	J Neurol. 264(1):102-111.	2017	3,41	
Neurodegenerative Diseases	**Travaglini L**, Aiello C, Alesi V, Loddo S, Novelli A, Tozzi G, Bertini E, Leuzzi V, Brancati F	Uniparental disomy of chromosome 1 unmasks recessive mutations of PPT1 in a boy with neuronal ceroid lipofuscinosis type 1	Brain Dev. 39(2):182-183.	2017	1,79	see METABOLIC
Seizures	**Balestri M**, Cappelletti S, Valeriani M, Vigevano F	Structural focal temporal lobe seizures in a child with lipoproteinosis	Pediatr Neurol.52(1):104-6.	2015	1,50	
Seizures	Barba C, Darra F, Cusmai R, Procopio E, Dionisi-Vici C, Keldermans L, Vuillaumier-Barrot S, Lefeber DJ, Guerrini R, CDG Group	Congenital disorders of glycosylation presenting as epileptic encephalopathy with migrating partial seizures in infancy	Dev Med Child Neurol. 58(10):1085-91.	2016	3,62	First and correspondig authors from University of Florence, see METABOLIC
Seizures	Battaglia A ,Bernardini L, Torrente I, Novelli A, Scarselli G	Spectrum of epilepsy and electroencephalogram patterns in idic (15) syndrome	Am J Med Genet A. 170(10):2531-9.	2016	2,08	First and correspondig authors from IRCCS Stella Maris, Pisa, see MALFORMATIONS AND ID
Seizures	**Bellusci M**, Trivisano M, De Palma L, Pietrafusa N, Vigevano F, Specchio N	Vigabatrin efficacy in GPR56-associated polymicrogyria: The role of GABAA receptor pathway	Epilepsia. 57(8):1337-8.	2016	4,71	

Seizures	**Cappelletti S**, Specchio N, Moavero R, Terracciano A, Pontrelli G, Gentile S, Vigevano F, Cusmai R	Cognitive development in females with PCDH19 gene-related epilepsy	Epilepsy Behav. 42:36-40.	2015	2,06	
Seizures	**Chiusolo F**, Diamanti A, Bianchi R, Fusco L, Elia M, Capriati T, Vigevano F, Picardo S	From intravenous to enteral ketogenic diet in PICU: A potential treatment strategy for refractory status epilepticus	Eur J Paediatr Neurol. 20(6):843-847.	2016	1,92	
Seizures	Cianchetti C, Vigevano F, Guidetti V	Epileptic headache: Need for a better knowledge	Headache. 55(4):580-1.	2015	2,76	First and correspondig authors from University of Cagliari
Seizures	**Compagnucci C**, Petrini S, Higuraschi N, Trivisano M, Specchio N, Hirose S, Bertini ES, Terracciano A	Characterizing PCDH19 in human induced pluripotent stem cells (iPSCs) and iPSC-derived developing neurons: Emerging role of a protein involved in controlling polarity during neurogenesis	Oncotarget. 6(29):26804-13.	2015	6,36	
Seizures	Coppola G, Besag F, Cusmai R, Dulac O, Kluger G, Moavero R, Nabbout R, Nikanorova M, Pisani F, Verrotti A, Von Stülpnagel C, Curatolo P	Current role of rufinamide in the treatment of childhood epilepsy: Literature review and treatment guidelines	Eur J Paediatr Neurol. 18(6):685-90.	2014	1,93	First and correspondig authors from other institutions
Seizures	Curatolo P, Aronica E, Jansen A, Jansen F, Kotulska K, Lagae L, **Moavero R**, Jozwiak S	Early onset epileptic encephalopathy or genetically determined encephalopathy with early onset epilepsy? Lessons learned from TSC	Eur J Paediatr Neurol. 20(2):203-11.	2016	2,30	

Seizures	**Cusmai R**, Verrotti A, Moavero R, Curatolo P, Battaglia D, Matricardi S, Spalice A, Vigevano F, Pruna D, Parisi P, D'Aniello A, Di Gennaro G, Coppola G	Rufinamide for the treatment of refractory epilepsy secondary to neuronal migration disorders	Epilepsy Res. 108(3):542-6.	2014	2,19	
Seizures	Granata T, Matricardi S, Ragona F, Freri E, Casazza M, Villani F, Deleo F, Tringali G, Gobbi G, Tassi L, Lo Russo G, Marras CE, Specchio N, Vigevano F, Fusco L	Hemispherotomy in Rasmussen encephalitis: Long-term outcome in an Italian series of 16 patients	Epilepsy Res. 108(6):1106-19.	2014	2,19	First and corresponding authors from Carlo Besta Neurological Institute, Milan
Seizures	Grosso S, Coppola G, Cusmai R, Parisi P, Spalice A, Foligno S, Verrotti A, Balestri P	Efficacy and tolerability of add-on lacosamide in children with Lennox-Gastaut syndrome	Acta Neurol Scand. 129(6):420-4.	2014	2,44	First and corresponding authors from University of Siena
Seizures	Mastrangelo M, Scheffer IE, Bramswig NC, Nair LD, Myers CT, Dentici ML, Korenke GC, Schoch K, Campeau PM, White SM, Shashi V, Kansagra S, Van Essen AJ, Leuzzi V	Epilepsy in KCNH1-related syndromes	Epileptic Disord. 18(2):123-36.	2016	0,94	First and correspondig authors from Sapienza University of Rome, see MALFORMATIONS AND ID

Seizures	Matricardi S, Spalice A, Salpietro V, Di Rosa G, Balistreri MC, Grosso S, Parisi P, Elia M, Striano P, Accorsi P, Cusmai R, Specchio N, Coppola G, Savasta S, Carotenuto M, Tozzi E, Ferrara P, Ruggieri M, Verrotti A	Epilepsy in the setting of full trisomy 18: A multicenter study on 18 affected children with and without structural brain abnormalities	Am J Med Genet C Semin Med Genet. 172(3):288-95	2016	4,47	First and correspondig authors from University of Chieti - Pescara, see MALFORMATIONS AND ID
Seizures	Nicita F, Spalice A, Raucci U, Iannetti P, Parisi P	The possible use of the L-type calcium channel antagonist verapamil in drug-resistant epilepsy	Expert Rev Neurother. 16(1):9-15.	2016	2,78	First and correspondig authors from Sapienza University of Rome
Seizures	**Pietrafusa N**, de Palma L, De Benedictis A, Trivisano M, Marras CE, Vigevano F, Specchio N	Ictal vomiting as a sign of temporal lobe epilepsy confirmed by stereo-EEG and surgical outcome	Epilepsy Behav. 53:112-6.	2015	2,26	
Seizures	**Pietrafusa N**, Trivisano M, de Palma L, Serino D, Moavero R, Benvenga A, Cappelletti S, Boero G, Vigevano F, La Neve A, Specchio N	Peri-ictal water drinking: a rare automatic behaviour in temporal lobe epilepsy	Epileptic Disord. 17(4):384-96.	2015	0,95	
Seizures	**Pontrelli G**, Cappelletti S, Claps D, Sirleto P, Ciocca L, Petrocchi S, Terracciano A, Serino D, Fusco L, Vigevano F, Specchio N	Epilepsy in patients with duplications of chromosome 14 harboring FOXG1	Pediatr Neurol. 50(5):530-5.	2014	1,50	see NEUROLOGICAL
Seizures	Serino D, Freri E, Ragona F, D'Incerti L, Bernardi B, Di Ciommo VM, Granata T, Vigevano F	Focal seizures versus epileptic spasms in children with focal cortical dysplasia and epilepsy onset in the first year	Epilepsy Res. 109:203-9.	2015	2,19	
Seizures	**Serino D**, Fusco L	Epileptic hypnagogic jerks mimicking repetitive sleep starts	Sleep Med. 16(8):1014-6.	2015	3,15	

Seizures	**Specchio N**, Pontrelli G, Serino D, Trivisano M, Cappelletti S, Terracciano A, Vigevano F, Fusco L	Occipital seizures induced by Intermittent Photic Stimulation in Dravet syndrome	Seizure. 23(4):309-13.	2014	2,06	
Seizures	**Specchio N**, Rizzi M, Fusco L, Rebessi E, Cappelletti S, De Palma L, Savioli A, De Benedictis A, Marras CE, Vigevano F, Delalande O	Acute intralesional recording in hypothalamic hamartoma: description of 4 cases	Acta Neurol Belg. 115(3):233-9.	2015	0,60	see PAEDIATRIC CANCERS
Seizures	**Terracciano A**, Trivisano M, Cusmai R, De Palma L, Fusco L, Compagnucci C, Bertini E, Vigevano F, Specchio N	PCDH19-related epilepsy in two mosaic male patients	Epilepsia. 57(3):e51-5.	2016	4,71	
Seizures	**Trivisano M**, Pietrafusa N, Ciommo Vd, Cappelletti S, Palma Ld, Terracciano A, Bertini E, Vigevano F, Specchio N	PCDH19-related epilepsy and Dravet Syndrome: Face-off between two early-onset epilepsies with fever sensitivity	Epilepsy Res. 125:32-6.	2016	2,24	
Seizures	**Trivisano M**, Specchio N, Vigevano F	Extending the use of stiripentol to other epileptic syndromes: A case of PCDH19-related epilepsy	Eur J Paediatr Neurol. 19(2):248-50.	2015	1,93	
Seizures	**Trivisano M**, Striano P, Sartorelli J, Giordano L, Traverso M, Accorsi P, Cappelletti S, Claps DJ, Vigevano F, Zara F, Specchio N	CHD2 mutations are a rare cause of generalized epilepsy with myoclonic-atonic seizures	Epilepsy Behav. 51:53-6.	2015	2,26	

Seizures	**Trivisano M**, Terracciano A, Milano T, Cappelletti S, Pietrafusa N, Bertini ES, Vigevano F, Specchio N	Mutation of CHRNA2 in a family with benign familial infantile seizures: Potential role of nicotinic acetylcholine receptor in various phenotypes of epilepsy	Epilepsia. 56(5):e53-7.	2015	4,57	
Seizures	Vecchi M, Barba C, De Carlo D, Stivala M, Guerrini R, Albamonte E, Ranalli D, Battaglia D, Lunardi G, Boniver C, Piccolo B, Pisani F, Cantalupo G, Nieddu G, Casellato S, Cappanera S, Cesaroni E, Zamponi N, Serino D, Fusco L, Iodice A, Palestra F, Giordano L, Freri E, De Giorgi I, Ragona F, Granata T, Fiocchi I, Bova SM, Mastrangelo M, Verrotti A, Matricardi S, Fontana E, Caputo D, Darra F, Dalla Bernardina B, Beccaria F, Capovilla G, Baglietto MP, Gagliardi A, Vignoli A, Canevini MP, Perissinotto E, Francione S	Symptomatic and presumed symptomatic focal epilepsies in childhood: An observational, prospective multicentre study	Epilepsia. 57(11):1808-1816.	2016	4,71	First and correspondig authors from University of Padua

Seizures	Verrotti A, Cusmai R, Laino D, Carotenuto M, Esposito M, Falsaperla R, Margari L, Rizzo R, Savasta S, Grosso S, Striano P, Belcastro V, Franzoni E, Curatolo P, Giordano L, Freri E, Matricardi S, Pruna D, Toldo I, Tozzi E, Lobefalo L, Operto F, Altobelli E, Chiarelli F, Spalice A	Long-term outcome of epilepsy in patients with Prader-Willi syndrome	J Neurol. 262(1):116-23.	2015	3,84	First and corresponding authors from University of Perugia, see MALFORMATIONS AND ID
Seizures	Verrotti A, Laino D, Rinaldi VE, Suppiej A, Giordano L, Toldo I, Margari L, Parisi P, Rizzo R, Matricardi S, Cusmai R, Grosso S, Gaggero R, Zamponi N, Pavone P, Capovilla G, Rauchenzauner M, Cerminara C, Di Gennaro G, Esposito M, Striano P, Savasta S, Coppola G, Siliquini S, Operto F, Belcastro V, Ragona F, Marseglia GL, Spalice A	Clinical dissection of childhood occipital epilepsy of Gastaut and prognostic implication	Eur J Neurol.23(2):241-6.	2016	4,06	First and correspondig authors from University of Perugia
Seizures	Villani F, Didato G, Deleo F, Tringali G, Garbelli R, Granata T, Marras CE, Cordella R, Spreafico R	Long-term outcome after limited cortical resections in two cases of adult-onset Rasmussen encephalitis	Epilepsia. 55(5):e38-43.	2014	4,58	First and corresponding authors from Carlo Besta Neurological Institute, Milan

Spastic paraplegia	Nicita F, Bertini E, Travaglini L, Armando M, Aiello C	Congenital-onset spastic paraplegia in a patient with TUBB4A mutation and mild hypomyelination	Neurol Sci. 368:145-6.	2016	2,13	First and correspondig authors from Sapienza University of Rome
Tuberous sclerosis	Curatolo P, Bjørnvold M, Dill PE, Ferreira JC, Feucht M, Hertzberg C, Jansen A, Jóźwiak S, Kingswood JC, Kotulska K, Macaya A, Moavero R, Nabbout R, Zonnenberg BA	The Role of mTOR Inhibitors in the Treatment of Patients with Tuberous Sclerosis Complex: Evidence-based and Expert Opinions	Drugs. 76(5):551-65.	2016	4,88	First and correspondig authors from Tor Vergata University of Rome, see MALFORMATIONS AND ID
Tuberous sclerosis	Curatolo P, Moavero R, de Vries P	Neurological and neuropsychiatric aspects of tuberous sclerosis complex	Lancet Neurol. 14(7):733-45.	2015	21,90	First and correspondig authors from Tor Vergata University of Rome, see MALFORMATIONS AND ID
Tuberous sclerosis	Curatolo P, Moavero R, Roberto D, Graziola F	Genotype/Phenotype Correlations in Tuberous Sclerosis Complex	Semin Pediatr Neurol. 22(4):259-73.	2015	2,23	First and correspondig authors from Tor Vergata University of Rome, see MALFORMATIONS AND ID
Tuberous sclerosis	Gialloreti LE, Moavero R, Marciano S, Pardini M, Benassi F, Mutolo MG, Curatolo P	Reduction in retinal nerve fiber layer thickness in tuberous sclerosis complex	Childs Nerv Syst. 31(6):857-61.	2015	1,11	First and correspondig authors from Tor Vergata University of Rome, see MALFORMATIONS AND ID

Tuberous Sclerosis	**Moavero R**, Folgiero V, Carai A, Miele E, Ferretti E, Po A, Diomedi Camassei F, Lepri FR, Vigevano F, Curatolo P, Valeriani M, Colafati GS, Locatelli F, Tornesello A, Mastronuzzi A	Metastatic Group 3 Medulloblastoma in a Patient With Tuberous Sclerosis Complex: Case Description and Molecular Characterization of the Tumor	Pediatr Blood Cancer. 63(4):719-22.	2016	2,39	see MALFORMATIONS AND ID
Tuberous Sclerosis	**Moavero R**, Napolitano A, Cusmai R, Vigevano F, Figà-Talamanca L, Calbi G, Curatolo P, Bernardi B	White matter disruption is associated with persistent seizures in tuberous sclerosis complex	Epilepsy Behav. 60:63-7.	2016	2,33	see MALFORMATIONS AND ID
Tuberous Sclerosis	**Moavero R**, Romagnoli G, Graziola F, Curatolo P	Mammalian Target of Rapamycin Inhibitors and Life-Threatening Conditions in Tuberous Sclerosis Complex	Semin Pediatr Neurol. 22(4):282-94.	2015	2,23	see MALFORMATIONS AND ID

Trial clinici farmacologici

***Trial* clinici per farmaci orfani per Malattie Rare dell'area specialistica con partecipazione dell'Unità operativa**

Centro di Eccellenza in Neuroscienze, Dipartimento NEURFARBA – Università degli Studi di Firenze, Azienda Ospedaliero-Universitaria A. Meyer, Firenze

Sono stati stimati circa > 27 Trials clinici in atto (20 su malattie neuromuscolari - coordinatore Mercuri). Trials preclinici sono in corso su PKU, disturbi del metabolismo della creatina.

Nel periodo compreso tra il 1 gennaio 2014 ed oggi, risultano in corso 5 trial clinici riguardanti il trattamento farmacologico dello stato di male epilettico refrattario e super-refrattario e di due sindromi epilettiche ad esordio esclusivamente pediatrico. Quattro sono i farmaci sperimentali, di cui la rufinamide, la fenfluramina e l'allopregnanolone hanno la designazione di farmaco orfano. Uno dei 5 studi è indipendente. Il Centro di Eccellenza in Neuroscienze dell'AOU Meyer è coordinatore di 3 dei 5 studi attivi.

Centro di Eccellenza in Neuroscienze, Dipartimento NEURFARBA – Università degli Studi di Firenze, Azienda Ospedaliero-Universitaria A. Meyer, Firenze

1 **Nome MR**	2 ***Sponsor***	3 ***Partners***		4 **Ruolo**		5 **Denominazione del *trial***	6 **Farmaco**	7 **Durata**		8 **Note**
		I	**N**	**C**	**P**			**C**	**T (anno)**	
SINDROME LENNOX–GASTAUT	EISAI	X			X	A multicenter, randomized,controlled,open - label study to evaluate the cognitive development effects and safety, and pharmacokinetics of adjunctive rufinamide treatment in pediatric subjects 1 to less than 4 years of age with inadequately controlled Lennox – Gastaut Syndrome	RUFINAMIDE		X (2015)	
SINDROME DI DRAVET	ZOGENIX	X		X		Sperimentazione multicentrica, randomizzata, in doppio cieco, a gruppi paralleli, controllata verso	FENFLURAMINA	X		

						placebo di due dosi fisse di soluzione orale ZX008 (fenfluramina cloridrato) come terapia supplementare in bambini e giovani adulti affetti dalla sindrome di Dravet				
SINDROME DI DRAVET	ZOGENIX	X		X		Studio di estensione in aperto per valutare la sicurezza a lungo termine di ZX008 (fenfluramina cloridrato) soluzione orale come terapia supplementare in bambini e adolescenti affetti dalla sindrome di Dravet.	FENFLURAMINA	X		
STATO EPILETTICO SUPER REFRATTARIO (SRSE)	SAGE THERAPEUTICS	X			X	Studio randomizzato, in doppio cieco e controllato con placebo, volto a valutare l'efficacia e la sicurezza dell'iniezione di SAGE-547 nel trattamento di pazienti affetti da stato di male epilettico super-refrattario (SRSE).	SAGE-547	X		SOLO LA PATOLOGIA E' DEFINITA RARA, COME RIPORTATO IN CTA FORM
STATO DI MALE EPILETTICO CONVULSIVO	AOU MEYER		X	X		Efficacia della ketamina nello stato di male epilettico convulsivo refrattario in età pediatrica: uno studio multicentrico, randomizzato, controllato, in aperto, no profit, con disegno sequenziale.	KETAMINA	X		n. 10 centri: ANCONA, BOLOGNA, FIRENZE, MILANO, PADOVA, ROMA (OPBG e GEMELLI) TORINO, TRIESTE, VERONA

Trials terapeutici in malattie neuromuscolari (NPI – Policlinico Gemelli – Roma) L

Syndrome	Study Code	Sponsor	Fase
DMD	H6D-MC-LVJJ	Eli Lilly	3
DMD	SRP4053-101	SAREPTA	I/II
DMD	PTC020E	PTC	3
SMA	ISIS396443-CS3B	ISIS	3
DMD	PRO044	Prosensa	2
SMA	ISIS396444-CS4	ISIS	3
SMA	BN29554	Roche	II/III
DMD	BMN-051-302	Biomarin Pharm. inc.	I/II
DMD	BMN-044-201	Biomarin Pharm. inc.	II
SMA	CS11	ISIS	
DMD	Natural History Study in Duchenne	Telethon	No Profit
SMA	BP39055	Roche	
SMA	BP39056	Roche	
DMD	B5161002	Pfizer	
DMD	PRO-DMD-01 Natural History	Prosensa (NL)	Osservaz.le
DMD	PRO045	Prosensa	2/3
DMD	PTC019	PTC	2
DMD	DSC11235743 Givino	Italfarmaco	2
DMD	SNT-[illegible] (SIDEROS)	Santhera	
DMD	PRO053	PROSENSA	I/II

Malattie reumatologiche pediatriche

A cura di REUMA (Gruppo di studio di Reumatologia Pediatrica della SIP)
(Coordinatore: *Angelo Ravelli*)

Marco Gattorno
Francesca Minoia
Angelo Ravelli
Alberto Tommasini

Pubblicazioni

Pubblicazioni del Gruppo di Studio di Reumatologia Pediatrica dal 2014 ad oggi

È stata effettuata una revisione delle principali pubblicazioni su riviste *peer reviwed* da parte dei componenti del Gruppo di Studio (Gds) di Reumatologia Pediatrica.

La ricerca è stata effettuata mediante PubMed, inserendo come intervallo di interesse il periodo dal 01/01/2014 al 16/06/2017 e restringendo la ricerca alle pubblicazioni originali, escludendo quindi i lavori di review e i case reports. Sono state incluse nell'elenco solo le pubblicazioni scritte o coordinate da un componente del Gds di Reumatologia Pediatrica.

Sono state quindi selezionate 89 pubblicazioni originali: la principale area di ricerca è risultata essere l'artrite idiopatica giovanile e le sue complicanze (uveite e sindrome da attivazione macrofagica), seguita in seconda istanza dalle patologie autoinfiammatorie.

Le ricerche sono state coordinate da 9 centri specialistici (Istituto G. Gaslini di Genova, Ospedale Pediatrico Bambino Gesù di Roma, Ospedale Pediatrico Meyer di Firenze, Istituto Burlo Garofalo di Trieste, Spedali Civili di Brescia, Azienda Ospedaliera di Padova, Istituto G. Pini di Milano, Ospedale dei Bambini di Palermo) e hanno complessivamente coinvolto 28 centri in tutta Italia.

Il 57% delle pubblicazioni ha in oggetto progetti multicentrici; il 68% coinvolge solo centri italiani, mentre il restante 32% ha previsto una collaborazione internazionale. Tra le pubblicazioni prevalgono le ricerche in ambito clinico (75%) rispetto quelle di laboratorio (25%).

La maggior parte degli articoli è stato pubblicato su riviste specializzate di Reumatologia e Immunologia (75%), ma circa il 20% è stato pubblicato su riviste non specialistiche, di cui 3 ad elevato imapct factor quali Lancet e JAMA.

Pubblicazioni

1 Nome MR	2 Autori	3 Titolo del lavoro	4 Rivista, volume, pagine	5 Anno	6 I. F.	7 Note
Uveitis	**Simonini G**, Bracaglia C, Cattalini M, Taddio A, Brambilla A, De Libero C, Pires Marafon D, Caputo R, Cimaz R	Predictors of Relapse after Discontinuing Systemic Treatment in Childhood Autoimmune Chronic Uveitis	J Rheumatol; 44:822-826	2017	3.24	
Arthritis	Favalli EG, Pontikaki I, Becciolini A, Biggioggero M, Ughi N, Romano M, Crotti C, Gattinara M, Gerloni V, Marchesoni A, Meroni PL	Real-life 10-year retention rate of first-line anti-TNF drugs for inflammatory arthritides in adult- and juvenile-onset populations: similarities and differences	Clin Rheumatol; [Epub ahead of print]	2017	2.04	
Autoinflammatory diseases	**Caorsi R**, Penco F, Grossi A, Insalaco A, Omenetti A, Alessio M, Conti G, Marchetti F, Picco P, Tommasini A, Martino S, Malattia C, Gallizi R, Podda RA, Salis A, Falcini F, Schena F, Garbarino F, Morreale A, Pardeo M, Ventrici C, Passarelli C, Zhou Q, Severino M, Gandolfo C, Damonte G, Martini A, Ravelli A, Aksentijevich I, Ceccherini I, **Gattorno M**	ADA2 deficiency (DADA2) as an unrecognised cause of early onset polyarteritis nodosa and stroke: a multicentre national study.	Ann Rheum Dis; [Epub ahead of print]	2017	12.38	
Primary angiitis of CNS	Rosati A, Cosi A, Basile M, Brambilla A, Guerrini R, Cimaz R, **Simonini G**	Mycophenolate mofetil as induction and long-term maintaining treatment in childhood: Primary angiitis of the central nervous system	Joint Bone Spine; 84:353-356	2017	2.94	
Juvenile idiopathic arthritis	Frid P, Nordal E, Bovis F, Giancane G, Larheim TA, Rygg M, Pires Marafon D, De Angelis D, Palmisani E, Murray KJ, Oliveira S, Simonini G, Corona F, Davidson J, Foster H, Steenks MH, Flato B, Zulian F, Baildam E, Saurenmann RK, Lahdenne P, Ravelli A, Martini A, Pistorio A, **Ruperto N;** Paediatric Rheumatology International Trials Organisation	Temporomandibular Joint Involvement in Association With Quality of Life, Disability, and High Disease Activity in Juvenile Idiopathic Arthritis	Arthritis Care Res; 69:677-686	2017	3.23	

Juvenile idiopathic arthritis	Cecchin V, Sperotto F, Balzarin M, Vittadello F, Martini G, **Zulian F**	Joint hypermobility and oligoarticular juvenile idiopathic arthritis: What relationship?	J Paediatr Child Health; 53:374-377	2017	1.47	
Juvenile idiopathic arthritis	**Ravelli A**, Davì S, Bracciolini G, Pistorio A, Consolaro A, van Dijkhuizen EH, Lattanzi B, Filocamo G, Verazza S, Gerloni V, Gattinara M, Pontikaki I, Insalaco A, De Benedetti F, Civino A, Presta G, Breda L, Marzetti V, Pastore S, Magni-Manzoni S, Maggio MC, Garofalo F, Rigante D, Gattorno M, Malattia C, Picco P, Viola S, Lanni S, Ruperto N, Martini A; Italian Pediatric Rheumatology Study Group	Intra-articular corticosteroids versus intra-articular corticosteroids plus methotrexate in oligoarticular juvenile idiopathic arthritis: a multicentre, prospective, randomised, open-label trial	Lancet; 389:909-916	2017	44.0	
Behcet disease	**Cantarini L**, Talarico R, Generali E, Emmi G, Lopalco G, Costa L, Silvestri E, Caso F, Franceschini R, Cimaz R, Iannone F, Galeazzi M, Selmi C	Safety profile of biologic agents for Behçet's disease in a multicenter observational cohort study	Int J Rheum Dis; 20:103-108	2017	1.91	
Macrophage activation syndrome	**Bracaglia C**, de Graaf K, Pires Marafon D, Guilhot F, Ferlin W, Prencipe G, Caiello I, Davì S, Schulert G, Ravelli A, Grom AA, de Min C, **De Benedetti F**	Elevated circulating levels of interferon-γ and interferon-γ-induced chemokines characterise patients with macrophage activation syndrome complicating systemic juvenile idiopathic arthritis	Ann Rheum Dis; 76:166-172	2017	12.38	
Kawasaki disease	**Taddio A**, Rossi ED, Monasta L, Pastore S, Tommasini A, Lepore L, Bronzetti G, Marrani E, Mottolese BD, Simonini G, Cimaz R, Ventura A	Describing Kawasaki shock syndrome: results from a retrospective study and literature review	Clin Rheumatol; 36:223-228	2017	2.04	

Juvenile idiopathic arthritis	Verazza S, Davì S, Consolaro A, Bovis F, Insalaco A, Magni-Manzoni S, Nicolai R, Marafon DP, De Benedetti F, Gerloni V, Pontikaki I, Rovelli F, Cimaz R, Marino A, Zulian F, Martini G, Pastore S, Sandrin C, Corona F, Torcoletti M, Conti G, Fede C, Barone P, Cattalini M, Cortis E, Breda L, Olivieri AN, Civino A, Podda R, Rigante D, La Torre F, D'Angelo G, Jorini M, Gallizzi R, Maggio MC, Consolini R, De Fanti A, Muratore V, Alpigiani MG, Ruperto N, Martini A, **Ravelli A**; Italian Pediatric Rheumatology Study Group	Disease status, reasons for discontinuation and adverse events in 1038 Italian children with juvenile idiopathic arthritis treated with etanercept	Pediatr Rheumatol Online J; 14:68	2016	2.14	
Juvenile idiopathic arthritis	Maggi L, Cosmi L, Simonini G, Annunziato F, **Cimaz R**	T cell subpopulations in juvenile idiopathic arthritis and their modifications after biotherapies	Autoimmun Rev; 15:1141-1144	2016	8.49	
Recurrent pericarditis	**Brucato A**, Imazio M, **Gattorno M**, Lazaros G, Maestroni S, Carraro M, Finetti M, Cumetti D, Carobbio A, Ruperto N, Marcolongo R, Lorini M, Rimini A, Valenti A, Erre GL, Sormani MP, Belli R, Gaita F, Martini A	Effect of Anakinra on Recurrent Pericarditis Among Patients With Colchicine Resistance and Corticosteroid Dependence: The AIRTRIP Randomized Clinical Trial	JAMA;316:1906-1912	2016	37.7	
Uveitis	Birolo C, Zannin ME, Arsenyeva S, Cimaz R, Miserocchi E, Dubko M, Deslandre CJ, Falcini F, Alessio M, La Torre F, Denisova E, Martini G, Nikishina I, **Zulian F**	Comparable Efficacy of Abatacept Used as First-line or Second-line Biological Agent for Severe Juvenile Idiopathic Arthritis-related Uveitis	J Rheumatol;43:2068-2073	2016	3.24	
Kawasaki disease	**Maggio MC**, Corsello G, Prinzi E, Cimaz R	Kawasaki disease in Sicily: clinical description and markers of disease severity	Ital J Pediatr;42:92	2016	1.61	

Arthritis	Vitale A, Insalaco A, Sfriso P, Lopalco G, Emmi G, Cattalini M, Manna R, Cimaz R, Priori R, Talarico R, Gentileschi S, de Marchi G, Frassi M, Gallizzi R, Soriano A, Alessio M, Cammelli D, Maggio MC, Marcolongo R, La Torre F, Fabiani C, Colafrancesco S, Ricci F, Galozzi P, Viapiana O, Verrecchia E, Pardeo M, Cerrito L, Cavallaro E, Olivieri AN, Paolazzi G, Vitiello G, Maier A, Silvestri E, Stagnaro C, Valesini G, Mosca M, de Vita S, Tincani A, Lapadula G, Frediani B, De Benedetti F, Iannone F, Punzi L, Salvarani C, Galeazzi M, Rigante D, **Cantarini L**	A Snapshot on the On-Label and Off-Label Use of the Interleukin-1 Inhibitors in Italy among Rheumatologists and Pediatric Rheumatologists: A Nationwide Multi-Center Retrospective Observational Study	Front Pharmacol;7:380	2016	4.41	
Arthritis	Margheri F, Ceruso M, Carta F, Laurenzana A, Maggi L, Lazzeri S, Simonini G, Annunziato F, Del Rosso M, Supuran CT, **Cimaz R**	Overexpression of the transmembrane carbonic anhydrase isoforms IX and XII in the inflamed synovium	J Enzyme Inhib Med Chem;31:60-63.	2016	3.42	
Autoinflammatory diseases	**Cattalini M**, Galli J, Andreoli L, Olivieri I, Ariaudo G, Fredi M; IAGSA study group, Orcesi S, Tincani A, Fazzi E.	Exploring Autoimmunity in a Cohort of Children with Genetically Confirmed Aicardi-Goutières Syndrome	J Clin Immunol;36:693-9	2016	3.09	
Juvenile idiopathic arthritis	Maggi L, Cimaz R, Capone M, Santarlasci V, Rossi MC, Mazzoni A, Montaini G, Pagnini I, Giani T, Simonini G, Scaletti C, Liotta F, Maggi E, Annunziato F, Cosmi L	Immunosuppressive Activity of Abatacept on Circulating T Helper Lymphocytes from Juvenile Idiopathic Arthritis Patients	Int Arch Allergy Immunol;171:45-53	2016	2.67	
Juvenile idiopathic arthritis	Di Paola M, Cavalieri D, Albanese D, Sordo M, Pindo M, Donati C, Pagnini I, Giani T, Simonini G, Paladini A, Lionetti P, De Filippo C, **Cimaz R**	Alteration of Fecal Microbiota Profiles in Juvenile Idiopathic Arthritis. Associations with HLA-B27 Allele and Disease Status	Front Microbiol;7:1703	2016	4.16	
Juvenile idiopathic arthritis	**Lanni S**, De Lucia O, Possemato N, Malattia C, Ravagnani V, Magni-Manzoni S	Musculoskeletal ultrasound in paediatric rheumatology: the Italian perspective	Clin Exp Rheumatol;34:957-958	2016	2.72	
Uveitis	Brambilla A, Caputo R, Cimaz R, **Simonini G**	Canakinumab for Childhood Sight-threatening Refractory Uveitis: A Case Series	J Rheumatol;43:1445-7	2016	3.24	

Autoinflammatory diseases	Pastore S, Ferrara G, Monasta L, Meini A, Cattalini M, Martino S, Alessio M, La Torre F, Teruzzi B, Gerloni V, Breda L, Taddio A, **Lepore L**	Chronic nonbacterial osteomyelitis may be associated with renal disease and bisphosphonates are a good option for the majority of patients	Acta Paediatr;105:e328-33	2016	1.64	
Juvenile idiopathic arthritis	Constantin T, Foeldvari I, Vojinovic J, Horneff G, Burgos-Vargas R, Nikishina I, Akikusa JD, Avcin T, Chaitow J, Koskova E, Lauwerys BR, Calvo Penades I, Flato B, Gamir ML, Huppertz HI, Jaller Raad JJ, Jarosova K, Anton J, Macku M, Otero Escalante WJ, Rutkowska-Sak L, Trauzeddel R, Velez-Sanchez PJ, Wouters C, Wajdula J, Zang C, Bukowski J, Woodworth D, Vlahos B, Martini A, **Ruperto N**; Paediatric Rheumatology International Trials Organisation (PRINTO).	Two-year Efficacy and Safety of Etanercept in Pediatric Patients with Extended Oligoarthritis, Enthesitis-related Arthritis, or Psoriatic Arthritis	J Rheumatol;43:816-24	2016	3.24	
Macrophage activation syndrome	**Ravelli A**, Minoia F, Davì S, Horne A, Bovis F, Pistorio A, Aricò M, Avcin T, Behrens EM, De Benedetti F, Filipovic L, Grom AA, Henter JI, Ilowite NT, Jordan MB, Khubchandani R, Kitoh T, Lehmberg K, Lovell DJ, Miettunen P, Nichols KE, Ozen S, Pachlopnik Schmid J, Ramanan AV, Russo R, Schneider R, Sterba G, Uziel Y, Wallace C, Wouters C, Wulffraat N, Demirkaya E, Brunner HI, Martini A, Ruperto N, Cron RQ	2016 Classification Criteria for Macrophage Activation Syndrome Complicating Systemic Juvenile Idiopathic Arthritis: A European League Against Rheumatism/American College of Rheumatology/Paediatric Rheumatology International Trials Organisation Collaborative Initiative	Ann Rheum Dis;75:481-9	2016	12.38	
Juvenile idiopathic arthritis	Nicolai R, Cortis E, Ravà L, Bracaglia C, Pardeo M, Insalaco A, Buonuomo PS, Tozzi AE, **De Benedetti F.**	Herpes Virus Infections During Treatment With Etanercept in Juvenile Idiopathic Arthritis	J Pediatric Infect Dis Soc;5:76-9	2016	0.83	

Macrophage activation syndrome	**Ravelli A**, Minoia F, Davì S, Horne A, Bovis F, Pistorio A, Aricò M, Avcin T, Behrens EM, De Benedetti F, Filipovic A, Grom AA, Henter JI, Ilowite NT, Jordan MB, Khubchandani R, Kitoh T, Lehmberg K, Lovell DJ, Miettunen P, Nichols KE, Ozen S, Pachlopnik Schmid J, Ramanan AV, Russo R, Schneider R, Sterba G, Uziel Y, Wallace C, Wouters C, Wulffraat N, Demirkaya E, Brunner HI, Martini A, Ruperto N, Cron RQ	Expert consensus on dynamics of laboratory tests for diagnosis of macrophage activation syndrome complicating systemic juvenile idiopathic arthritis	RMD Open;2:e000161	2016		
Juvenile idiopathic arthritis	**Lanni S**, Bovis F, Ravelli A, Viola S, Magnaguagno F, Pistorio A, Michele Magnano G, Martini A, **Malattia C**	Delineating the Application of Ultrasound in Detecting Synovial Abnormalities of the Subtalar Joint in Juvenile Idiopathic Arthritis	Arthritis Care Res;68:1346-53	2016	3.23	
Juvenile idiopathic arthritis	Demirkaya E, Lanni S, Bovis F, Galasso R, Ravelli A, Palmisani E, Consolaro A, Pederzoli S, Marafon D, Simianer S, Martini A, **Ruperto N**, Pistorio A; Paediatric Rheumatology International Trials Organisation	A Meta-Analysis to Estimate the Placebo Effect in Randomized Controlled Trials in Juvenile Idiopathic Arthritis	Arthritis Rheumatol;68:1540-50	2016	8.95	
Uveitis	Miserocchi E, Modorati G, Berchicci L, Pontikaki I, Meroni P, **Gerloni V**	Long-term treatment with rituximab in severe juvenile idiopathic arthritis-associated uveitis	Br J Ophthalmol;100:782-6	2016	3.03	
Juvenile idiopathic arthritis	Maggi L, Margheri F, Luciani C, Capone M, Rossi MC, Chillà A, Santarlasci V, Mazzoni A, Cimaz R, Liotta F, Maggi E, Cosmi L, Del Rosso M, Annunziato F	Th1-Induced CD106 Expression Mediates Leukocytes Adhesion on Synovial Fibroblasts from Juvenile Idiopathic Arthritis Patients	PLoS One;11:e0154422	2016	3.23	
Kawasaki disease	Mauro A, Fabi M, Da Frè M, Guastaroba P, Corinaldesi E, Calabri GB, Giani T, Simonini G, Rusconi F, **Cimaz R**	Kawasaki disease: an epidemiological study in central Italy	Pediatr Rheumatol Online J;14:22	2016	2.14	

Juvenile dermatomyositis	**Ruperto N**, Pistorio A, Oliveira S, Zulian F, Cuttica R, Ravelli A, Fischbach M, Magnusson B, Sterba G, Avcin T, Brochard K, Corona F, Dressler F, Gerloni V, Apaz MT, Bracaglia C, Cespedes-Cruz A, Cimaz R, Couillault G, Joos R, Quartier P, Russo R, Tardieu M, Wulffraat N, Bica B, Dolezalova P, Ferriani V, Flato B, Bernard-Medina AG, Herlin T, Trachana M, Meini A, Allain-Launay E, Pilkington C, Vargova V, Wouters C, Angioloni S, Martini A; Paediatric Rheumatology International Trials Organisation (PRINTO).	Prednisone versus prednisone plus ciclosporin versus prednisone plus methotrexate in new-onset juvenile dermatomyositis: a randomised trial	Lancet;387:671-8	2016	44.0	
Arthritis	**Cattalini M**, Parissenti I, Tononcelli E, Lancini F, Cantarini L, Meini A	Developing a Predictive Score for Chronic Arthritis among a Cohort of Children with Musculoskeletal Complaints--The Chronic Arthritis Score Study	J Pediatr;169:188-93	2016	3.89	
Macrophage activation syndrome	**Ravelli A**, Minoia F, Davì S, Horne A, Bovis F, Pistorio A, Aricò M, Avcin T, Behrens EM, De Benedetti F, Filipovic L, Grom AA, Henter JI, Ilowite NT, Jordan MB, Khubchandani R, Kitoh T, Lehmberg K, Lovell DJ, Miettunen P, Nichols KE, Ozen S, Pachlopnik Schmid J, Ramanan AV, Russo R, Schneider R, Sterba G, Uziel Y, Wallace C, Wouters C, Wulffraat N, Demirkaya E, Brunner HI, Martini A, Ruperto N, Cron RQ	2016 Classification Criteria for Macrophage Activation Syndrome Complicating Systemic Juvenile Idiopathic Arthritis: A European League Against Rheumatism/American College of Rheumatology/Paediatric Rheumatology International Trials Organisation Collaborative Initiative	Arthritis Rheumatol;68:566-76	2016	8.95	
Juvenile idiopathic arthritis	Colebatch-Bourn AN, Edwards CJ, Collado P, D'Agostino MA, Hemke R, Jousse-Joulin S, Maas M, Martini A, Naredo E, Østergaard M, Rooney M, Tzaribachev N, van Rossum MA, Vojinovic J, Conaghan PG, **Malattia C**	EULAR-PReS points to consider for the use of imaging in the diagnosis and management of juvenile idiopathic arthritis in clinical practice	Ann Rheum Dis;74:1946-57	2015	12.38	

Macrophage activation syndrome	Cifaldi L, Prencipe G, Caiello I, Bracaglia C, Locatelli F, **De Benedetti F**, Strippoli R	Inhibition of natural killer cell cytotoxicity by interleukin-6: implications for the pathogenesis of macrophage activation syndrome	Arthritis Rheumatol;67:3037-46	2015	8.95	
Juvenile idiopathic arthritis	Piancino MG, Cannavale R, Dalmasso P, Tonni I, Filipello F, Perillo L, Cattalini M, **Meini A**	Condylar asymmetry in patients with juvenile idiopathic arthritis: Could it be a sign of a possible temporomandibular joints involvement?	Semin Arthritis Rheum;45:208-13	2015	3.94	
Uveitis	Marrani E, Cimaz R, Lucherini OM, Caputo R, Vitale A, Cantarini L, **Simonini G**	The common NOD2/CARD15 variant P268S in patients with non-infectious uveitis: a cohort study	Pediatr Rheumatol Online J;13:38	2015	2.14	
Autoinflammatory diseases	**Cattalini M**, Soliani M, Rigante D, Lopalco G, Iannone F, Galeazzi M, Cantarini L	Basic Characteristics of Adults with Periodic Fever, Aphthous Stomatitis, Pharyngitis, and Adenopathy Syndrome in Comparison with the Typical Pediatric Expression of Disease	Mediators Inflamm;2015:570418	2015	3.41	
Uveitis	Marrani E, Paganelli V, de Libero C, Cimaz R, **Simonini G**	Long-term efficacy of abatacept in pediatric patients with idiopathic uveitis: a case series.	Graefes Arch Clin Exp Ophthalmol;253:1813-6	2015	1.99	
Juvenile idiopathic arthritis	Real-Fernández F, Cimaz R, Rossi G, Simonini G, Giani T, Pagnini I, Papini AM, Rovero P	Surface plasmon resonance-based methodology for anti-adalimumab antibody identification and kinetic characterization	Anal Bioanal Chem;407:7477-85	2015	3.12	
Juvenile idiopathic arthritis	Pardeo M, Pires Marafon D, Insalaco A, Bracaglia C, Nicolai R, Messia V, **De Benedetti F**	Anakinra in Systemic Juvenile Idiopathic Arthritis: A Single-center Experience	J Rheumatol;42:1523-7	2015	3.24	
Behcet disease	**Cantarini** L, Vitale A, Scalini P, Dinarello CA, Rigante D, Franceschini R, Simonini G, Borsari G, Caso F, Lucherini OM, Frediani B, Bertoldi I, Punzi L, Galeazzi M, Cimaz R. Cantarini L, Vitale A, Scalini P, Dinarello CA, Rigante D, Franceschini R, Simonini G, Borsari G, Caso F, Lucherini OM, Frediani B, Bertoldi I, Punzi L, Galeazzi M, **Cimaz R**	Anakinra treatment in drug-resistant Behcet's disease: a case series	Clin Rheumatol;34:1293-301	2015	2.04	

Macrophage activation syndrome	**Minoia F**, Davì S, Horne A, Bovis F, Demirkaya E, Akikusa J, Ayaz NA, Al-Mayouf SM, Barone P, Bica B, Bolt I, Breda L, De Cunto C, Enciso S, Gallizzi R, Griffin T, Hennon T, Horneff G, Jeng M, Kapovic AM, Lipton JM, Magni Manzoni S, Rumba-Rozenfelde I, Magalhaes CS, Sewairi WM, Stine KC, Vougiouka O, Weaver LK, Davidsone Z, De Inocencio J, Ioseliani M, Lattanzi B, Tezer H, Buoncompagni A, Picco P, Ruperto N, Martini A, Cron RQ, **Ravelli A**	Dissecting the heterogeneity of macrophage activation syndrome complicating systemic juvenile idiopathic arthritis	J Rheumatol;42:994-1001	2015	3.24	
Autoinflammatory diseases	Carta S, Penco F, Lavieri R, Martini A, Dinarello CA, **Gattorno M**, Rubartelli A	Cell stress increases ATP release in NLRP3 inflammasome-mediated autoinflammatory diseases, resulting in cytokine imbalance	Proc Natl Acad Sci U S A;112:2835-40	2015	9.67	
Autoinflammatory diseases	De Pieri C, Taddio A, Insalaco A, Barbi E, Lepore L, Ventura A, **Tommasini A**	Different presentations of mevalonate kinase deficiency: a case series	Clin Exp Rheumatol;33:437-42	2015	2.72	
Arthritis	Sperotto F, Brachi S, Vittadello F, **Zulian F**.	Musculoskeletal pain in schoolchildren across puberty: a 3-year follow-up study	Pediatr Rheumatol Online J;13:16	2015	2.14	
Juvenile idiopathic arthritis	**Pastore S**, Stocco G, Moressa V, Zandonà L, Favretto D, Malusà N, Decorti G, **Lepore L**, Ventura A	5-Aminoimidazole-4-carboxamide ribonucleotide-transformylase and inosine-triphosphate-pyrophosphatase genes variants predict remission rate during methotrexate therapy in patients with juvenile idiopathic arthritis	Rheumatol Int;35:619-27	2015	1.70	
Juvenile idiopathic arthritis	**Martini G**, Biscaro F, Boscaro E, Calabrese F, Lunardi F, Facco M, Agostini C, Zulian F, Fadini GP	Reduced levels of circulating progenitor cells in juvenile idiopathic arthritis are counteracted by anti TNF-α therapy	BMC Musculoskelet Disord;16:103	2015	1.68	

Juvenile idiopathic arthritis	**De Benedetti F**, Brunner H, Ruperto N, Schneider R, Xavier R, Allen R, Brown DE, Chaitow J, Pardeo M, Espada G, Gerloni V, Myones BL, Frane JW, Wang J, Lipman TH, Bharucha KN, Martini A, Lovell D; Paediatric Rheumatology International Trials Organisation and the Pediatric Rheumatology Collaborative Study Group	Catch-up growth during tocilizumab therapy for systemic juvenile idiopathic arthritis: results from a phase III trial.	Arthritis Rheumatol;67:840-8	2015	8.95	
Juvenile idiopathic arthritis	**Pastore S**, Stocco G, Favretto D, De Iudicibus S, Taddio A, d'Adamo P, Malusà N, Addobbati R, Decorti G, Lepore **L**, Ventura A	Genetic determinants for methotrexate response in juvenile idiopathic arthritis	Front Pharmacol;6:52	2015	4.41	
Autoinflammatory diseases	De Pieri C, Vuch J, De Martino E, Bianco AM, Ronfani L, Athanasakis E, Bortot B, Crovella S, Taddio A, Severini GM, **Tommasini A**	Genetic profiling of autoinflammatory disorders in patients with periodic fever: a prospective study.	Pediatr Rheumatol Online J;13:11	2015	2.14	
Connective tissue disease	**Falcini F**, Rigante D, Candelli M, Martini G, Corona F, Petaccia A, La Torre F, Raffaele CG, Matucci Cerinic M	Anti-nuclear antibodies as predictor of outcome in a multi-center cohort of Italian children and adolescents with Raynaud's phenomenon	Clin Rheumatol;34:167-9	2015	2.04	
Autoinflammatory diseases	De Pieri C, Vuch J, Athanasakis E, Severini GM, Crovella S, Bianco AM, **Tommasini A.**	F402L variant in NLRP12 in subjects with undiagnosed periodic fevers and in healthy controls	Clin Exp Rheumatol;32:993-4	2014	2.72	
Macrophage activation syndrome	**Minoia F**, Davì S, Horne A, Demirkaya E, Bovis F, Li C, Lehmberg K, Weitzman S, Insalaco A, Wouters C, Shenoi S, Espada G, Ozen S, Anton J, Khubchandani R, Russo R, Pal P, Kasapcopur O, Miettunen P, Maritsi D, Merino R, Shakoory B, Alessio M, Chasnyk V, Sanner H, Gao YJ, Huasong Z, Kitoh T, Avcin T, Fischbach M, Frosch M, Grom A, Huber A, Jelusic M, Sawhney S, Uziel Y, Ruperto N, Martini A, Cron RQ, **Ravelli A**	Clinical features, treatment, and outcome of macrophage activation syndrome complicating systemic juvenile idiopathic arthritis: a multinational, multicenter study of 362 patients	Arthritis Rheumatol;66:3160-9	2014	8.95	

Macrophage activation syndrome	**Davì S**, Minoia F, Pistorio A, Horne A, Consolaro A, Rosina S, Bovis F, Cimaz R, Gamir ML, Ilowite NT, Kone-Paut I, Feitosa de Oliveira SK, McCurdy D, Silva CA, Sztajnbok F, Tsitsami E, Unsal E, Weiss JE, Wulffraat N, Abinun M, Aggarwal A, Apaz MT, Astigarraga I, Corona F, Cuttica R, D'Angelo G, Eisenstein EM, Hashad S, Lepore L, Mulaosmanovic V, Nielsen S, Prahalad S, Rigante D, Stanevicha V, Sterba G, Susic G, Takei S, Trauzeddel R, Zletni M, Ruperto N, Martini A, Cron RQ, **Ravelli A**	Performance of current guidelines for diagnosis of macrophage activation syndrome complicating systemic juvenile idiopathic arthritis.	Arthritis Rheumatol;66:2871-80	2014	8.95	
Juvenile idiopathic arthritis	**Consolaro A**, Negro G, Chiara Gallo M, Bracciolini G, Ferrari C, Schiappapietra B, Pistorio A, Bovis F, Ruperto N, Martini A, **Ravelli A**	Defining criteria for disease activity states in nonsystemic juvenile idiopathic arthritis based on a three-variable juvenile arthritis disease activity score	Arthritis Care Res;66:1703-9	2014	3.23	
Juvenile idiopathic arthritis	Caiello I, Minnone G, Holzinger D, Vogl T, Prencipe G, Manzo A, **De Benedetti F**, Strippoli R	IL-6 amplifies TLR mediated cytokine and chemokine production: implications for the pathogenesis of rheumatic inflammatory diseases	PLoS One;9:e107886	2014	3.23	
Autoinflammatory diseases	**Pastore S**, Paloni G, Caorsi R, Ronfani L, Taddio A, **Lepore L;** CAPS Italian Register	Serum amyloid protein A concentration in cryopyrin-associated periodic syndromes patients treated with interleukin-1 beta antagonist	Clin Exp Rheumatol;32:S63-6	2014	2.72	
Autoinflammatory diseases	Vigo G, Martini G, Zoppi S, Vittadello F, **Zulian F**.	Tonsillectomy efficacy in children with PFAPA syndrome is comparable to the standard medical treatment: a long-term observational study	Clin Exp Rheumatol;32:S156-9	2014	2.72	
Juvenile idiopathic arthritis	Calandra S, Gallo MC, Consolaro A, Pistorio A, Lattanzi B, Bovis F, Muratore V, De Marco R, Martini A, **Ravelli A**	Female sex and oligoarthritis category are not risk factors for uveitis in Italian children with juvenile idiopathic arthritis.	J Rheumatol;41:1416-25	2014	3.24	

Juvenile idiopathic arthritis	Sperotto F, Cuffaro G, Brachi S, Seguso M, **Zulian F**	Prevalence of antinuclear antibodies in schoolchildren during puberty and possible relationship with musculoskeletal pain: a longitudinal study	J Rheumatol;41:1405-8.	2014	3.24	
Juvenile idiopathic arthritis	**Consolaro A**, Ruperto N, Bracciolini G, Frisina A, Gallo MC, Pistorio A, Verazza S, Negro G, Gerloni V, Goldenstein-Schainberg C, Sztajnbok F, Wulffraat NM, Martini A, **Ravelli A**; Paediatric Rheumatology International Trials Organization (PRINTO)	Defining criteria for high disease activity in juvenile idiopathic arthritis based on the juvenile arthritis disease activity score	Ann Rheum Dis;73:1380-3	2014	12.38	
Autoinflammatory diseases	Scianaro R, Insalaco A, Bracci Laudiero L, De Vito R, Pezzullo M, Teti A, De Benedetti F, **Prencipe G**	Deregulation of the IL-1β axis in chronic recurrent multifocal osteomyelitis	Pediatr Rheumatol Online J;12:30	2014	2.14	
Juvenile dermatomyositis	**Malattia C**, Damasio MB, Madeo A, Pistorio A, Providenti A, Pederzoli S, Viola S, Buoncompagni A, Mattiuz C, Beltramo A, Consolaro A, Ravelli A, Ruperto N, Picco P, Magnano GM, Martini A	Whole-body MRI in the assessment of disease activity in juvenile dermatomyositis.	Ann Rheum Dis;73:1083-90	2014	12.38	
Juvenile idiopathic arthritis	Brunner HI, Ruperto N, Zuber Z, Keane C, Harari O, Kenwright A, Lu P, Cuttica R, Keltsev V, Xavier RM, Calvo I, Nikishina I, Rubio-Pérez N, Alexeeva E, Chasnyk V, Horneff G, Opoka-Winiarska V, Quartier P, Silva CA, Silverman E, Spindler A, Baildam E, Gámir ML, Martin A, Rietschel C, Siri D, Smolewska E, Lovell D, Martini A, **De Benedetti F**; Paediatric Rheumatology International Trials Organisation PRINTO; Pediatric Rheumatology Collaborative Study Group (PRCSG)	Efficacy and safety of tocilizumab in patients with polyarticular-course juvenile idiopathic arthritis: results from a phase 3, randomised, double-blind withdrawal trial	Ann Rheum Dis;74:1110-7	2014	12.38	

Polyarteritis nodosa	Falcini F, La Torre F, Vittadello F, Rigante D, Martini G, Corona F, Buoncompagni A, Alessio M, Cortis E, Insalaco A, Magni-Manzoni S, Breda L, Matucci-Cerinic M, **Zulian F**	Clinical overview and outcome in a cohort of children with polyarteritis nodosa	Clin Exp Rheumatol;32:S134-7	2014	2.72	
Granulomatosis with polyangiitis	Bohm M, Gonzalez Fernandez MI, Ozen S, Pistorio A, Dolezalova P, Brogan P, Barbano G, Sengler C, Klein-Gitelman M, Quartier P, Fasth A, Herlin T, Terreri MT, Nielsen S, van Rossum MA, Avcin T, Castell ER, Foeldvari I, Foell D, Kondi A, Koné-Paut I, Kuester RM, Michels H, Wulffraat N, Amer HB, Malattia C, Martini A, **Ruperto N**	Clinical features of childhood granulomatosis with polyangiitis (wegener's granulomatosis)	Pediatr Rheumatol Online J;12:18	2014	2.14	
Juvenile idiopathic arthritis	**Magni-Manzoni S**, Collado P, Jousse-Joulin S, Naredo E, D'Agostino MA, Muratore V, Merli P, Roth J; Paediatric Ultrasound Group of the OMERACT Ultrasound Task Force.	Current state of musculoskeletal ultrasound in paediatric rheumatology: results of an international survey	Rheumatology;53:491-6	2014	4.52	
Juvenile idiopathic arthritis	Romano M, Pontikaki I, Gattinara M, Ardoino I, Donati C, Boracchi P, Meroni PL, **Gerloni V**	Drug survival and reasons for discontinuation of the first course of biological therapy in 301 juvenile idiopathic arthritis patients	Reumatismo;65:278-85	2014	4.52	
Camptodactyly-arthropathy-coxa vara-pericarditis	Ciullini Mannurita S, Vignoli M, Bianchi L, Kondi A, Gerloni V, Breda L, Ten Cate R, Alessio M, Ravelli A, Falcini F, Gambineri E	CACP syndrome: identification of five novel mutations and of the first case of UPD in the largest European cohort	Eur J Hum Genet;22:197-201	2014	4.58	
Juvenile idiopathic arthritis	Del Fattore A, Cappariello A, Capulli M, Rucci N, Muraca M, De Benedetti F, Teti A	An experimental therapy to improve skeletal growth and prevent bone loss in a mouse model overexpressing IL-6	Osteoporos Int;25:681-92	2014	3.44	
Juvenile idiopathic arthritis	**Giancane G**, Pederzoli S, Norambuena X, Ioseliani M, Sato J, Gallo MC, Negro G, Pistorio A, Ruperto N, Martini A, **Ravelli A**	Frequency of radiographic damage and progression in individual joints in children with juvenile idiopathic arthritis	Arthritis Care Res;66:27-33	2014	3.23	

Juvenile idiopathic arthritis	Rodriguez-Lozano AL, Giancane G, Pignataro R, Viola S, Valle M, Gregorio S, Norambuena X, Ioseliani M, Pistorio A, Magnaguagno F, Riganti S, Martini A, **Ravelli A**	Agreement among musculoskeletal pediatric specialists in the assessment of radiographic joint damage in juvenile idiopathic arthritis	Arthritis Care Res;66:34-9	2014	3.23	
Autoinflammatory syndromes	**Gattorno M**, Obici L, Cattalini M, Tormey V, Abrams K, Davis N, Speziale A, Bhansali SG, Martini A, Lachmann HJ.	Canakinumab treatment for patients with active recurrent or chronic TNF receptor-associated periodic syndrome (TRAPS): an open-label, phase II study	Ann Rheum Dis. 76(1):173-178	2017	12,32	Gaslini Genova
Autoinflammatory syndromes	Torene R, Nirmala N, Obici L, Cattalini M, Tormey V, Caorsi R, Starck-Schwertz S, Letzkus M, Hartmann N, Abrams K, Lachmann H, **Gattorno M**	Canakinumab reverses overexpression of inflammatory response genes in tumour necrosis factor receptor-associated periodic syndrome.	Ann Rheum Dis. 76(1):303-309	2017	12,32	Gaslini Genova
Autoinflammatory syndromes	**Finetti M**, Omenetti A, Federici S, Caorsi R, **Gattorno M**	Chronic Infantile Neurological Cutaneous and Articular (CINCA) syndrome: a review	Orphanet J Rare Dis. 7;11(1):167	2016		Gaslini Genova
Autoinflammatory syndromes	Ter Haar NM, Jeyaratnam J, Lachmann HJ, Simon A, Brogan PA, Doglio M, Cattalini M, Anton J, Modesto C, Quartier P, Hoppenreijs E, Martino S, Insalaco A, Cantarini L, Lepore L, Alessio M, Calvo Penades I, Boros C, Consolini R, Rigante D, Russo R, Pachlopnik Schmid J, Lane T, Martini A, Ruperto N, Frenkel J, and **Gattorno M;** Paediatric Rheumatology International Trials Organisation and Eurofever Project.	The Phenotype and Genotype of Mevalonate Kinase Deficiency: A Series of 114 Cases From the Eurofever Registry	Arthritis Rheumatol. 68(11):2795-2805	2016	6	Gaslini Genova
Autoinflammatory syndromes	Marsili M, Di Battista C, Chiarelli F, Breda L, Ceccherini I, **Gattorno M**	Neonatal-Onset Urticaria and Fever	J Pediatr.177:329-329	2016	3,89	Gaslini Genova
Autoinflammatory syndromes	Omenetti A, Carta S, Caorsi R, Finetti M, Marotto D, Lattanzi B, Jorini M, Delfino	Disease activity accounts for long-term efficacy of IL-1 blockers in pyogenic sterile	Rheumatology (Oxford). 55(7):1325-35	2016	4,52	Gaslini Genova

	L, Penco F, Picco P, Buoncompagni A, Martini A, Rubartelli A, **Gattorno M**	arthritis pyoderma gangrenosum and severe acne syndrome				
Autoinflammatory syndromes	**Caorsi R**, Penco F, Schena F, **Gattorno M**	Monogenic polyarteritis: the lesson of ADA2 deficiency	Pediatr Rheumatol Online J. 14(1):51	2016	2,14	Gaslini Genova
Autoinflammatory syndromes	Volpi S, Picco P, Caorsi R, Candotti F, **Gattorno M**	Type I interferonopathies in pediatric rheumatology	Pediatr Rheumatol Online J. 14(1):35	2016	2,14	Gaslini Genova
Autoinflammatory syndromes	Imazio M, Brucato A, Pluymaekers N, Breda L, Calabri G, Cantarini L, Cimaz R, Colimodio F, Corona F, Cumetti D, Cuccio CD, Gattorno M, Insalaco A, Limongelli G, Russo MG, Valenti A, Finkelstein Y, **Martini A**	Recurrent pericarditis in children and adolescents: a multicentre cohort study.	J Cardiovasc Med (Hagerstown). 17(9):707-12	2016		Gaslini Genova
Autoinflammatory syndromes	**Papa R**, Nozza P, Granata C, Caorsi R, Gattorno M, Martini A, **Picco P.**	Juvenile eosinophilic fasciitis: three case reports with review of the literature.	Clin Exp Rheumatol. 34(3):527-30	2016	2,49	Gaslini Genova
Autoinflammatory syndromes	**Naselli A**, Penco F, Cantarini L, Insalaco A, Alessio M, Tommasini A, Maggio C, Obici L, Gallizi R, Cimmino M, Signa S, Lucherini OM, Carta S, Caroli F, Martini A, Rubartelli A, Ceccherini I, **Gattorno M**	Clinical Characteristics of Patients Carrying the Q703K Variant of the NLRP3 Gene: A 10-year Multicentric National Study	J Rheumatol. 43(6):1093-100	2016	3,23	Gaslini Genova
Autoinflammatory syndromes	Demirkaya E, Saglam C, Turker T, Koné-Paut I, Woo P, Doglio M, Amaryan G, Frenkel J, Uziel Y, Insalaco A, Cantarini L, Hofer M, Boiu S, Duzova A, Modesto C, Bryant A, Rigante D, Papadopoulou-Alataki E, Guillaume-Czitrom S, Kuemmerle-Deschner J, Neven B, Lachmann H, Martini A, Ruperto N, **Gattorno M***, Ozen S*; Paediatric Rheumatology International Trials	Performance of Different Diagnostic Criteria for Familial Mediterranean Fever in Children with Periodic Fevers: Results from a Multicenter International Registry.	J Rheumatol. 43(1):154-60	2016	3,23	Gaslini Genova

	Organisations (PRINTO).; Eurofever Project. (*last co-authors)					
Autoinflammatory syndromes	Rusmini M, Federici S, Caroli F, Grossi A, Baldi M, Obici L, Insalaco A, Tommasini A, Caorsi R, Gallo E, Olivieri AN, Marzano A, Coviello D, Ravazzolo R, Martini A, **Gattorno* M**, Ceccherini I*. (*last co-authors)	Next-generation sequencing and its initial applications for molecular diagnosis of systemic auto-inflammatory diseases.	Ann Rheum Dis. pii: annrheumdis-2015-207701	2015	12,32	Gaslini Genova
Autoinflammatory syndromes	Holzinger D, Kessel C, Omenetti A, **Gattorno M**.	From bench to bedside and back again: translational research in autoinflammation.	Nat Rev Rheumatol. doi: 10.1038/nrrheum.2015.79	2015	10,53	Gaslini Genova
Autoinflammatory syndromes	Ter Haar NM, Oswald M, Jeyaratnam J, Anton J, Barron KS, Brogan PA, Cantarini L, Galeotti C, Grateau G, Hentgen V, Hofer M, Kallinich T, Kone-Paut I, Lachmann HJ, Ozdogan H, Ozen S, Russo R, Simon A, Uziel Y, Wouters C, Feldman BM, Vastert SJ, Wulffraat NM, Benseler SM, Frenkel J, **Gattorno M***, Kuemmerle-Deschner JB*.	Recommendations for the management of autoinflammatory diseases.	Ann Rheum Dis. 274(9):1636-44. (IF 12.32)	2015	12,32	Gaslini Genova
Autoinflammatory syndromes	**Federici S**, Sormani MP, Ozen S, Lachmann HJ, Amaryan G, Woo P, Koné-Paut I, Dewarrat N, Cantarini L, Insalaco A, Uziel Y, Rigante D, Quartier P, Demirkaya E, Herlin T, Meini A, Fabio G, Kallinich T, Martino S, Butbul AY, Olivieri A, Kuemmerle-Deschner J, Neven B, Simon A, Ozdogan H, Touitou I, Frenkel J, Hofer M, Martini A, Ruperto N,	Evidence-based provisional clinical classification criteria for autoinflammatory periodic fevers	nn Rheum Dis. 74(5):799-805. (IF 12.32)	2015	12,32	Gaslini Genova

	Gattorno M; for the Paediatric Rheumatology International Trials Organisation (PRINTO) and Eurofever Project.					
Autoinflammatory syndromes	Levy R, Gérard L, Kuemmerle-Deschner J, Lachmann HJ, Koné-Paut I, Cantarini L, Woo P, Naselli A, Bader-Meunier B, Insalaco A, Al-Mayouf SM, Ozen S, Hofer M, Frenkel J, Modesto C, Nikishina I, Schwarz T, Martino S, Meini A, Quartier P, Martini A, Ruperto N, Neven B, **Gattorno M**; for PRINTO and Eurofever	Phenotypic and genotypic characteristics of cryopyrin-associated periodic syndrome: a series of 136 patients from the Eurofever Registry	Ann Rheum Dis. 74(11):2043-9	2015	12,32	Gaslini Genova
Autoinflammatory syndromes	Marzano AV, Tavecchio S, Venturini M, Sala R, Calzavara-Pinton P, **Gattorno M.**	Urticarial vasculitis and urticarial autoinflammatory syndromes.	G Ital Dermatol Venereol. 150(1):41-50c	2015	1,01	Gaslini Genova
Autoinflammatory syndromes	Lachmann HJ, Papa R, Gerhold K, Obici L, Touitou I, Cantarini L, Frenkel J, Anton J, Kone-Paut I, Cattalini M, Bader-Meunier B, Insalaco A, Hentgen V, Merino R, Modesto C, Toplak N, Berendes R, Ozen S, Cimaz R, Jansson A, Brogan PA, Hawkins PN, Ruperto N, Martini A, Woo P and **Gattorno M**; for the Paediatric Rheumatology International Trials Organisation (PRINTO), the EUROTRAPS and the Eurofever Project	The phenotype of TNF receptor-associated autoinflammatory syndrome (TRAPS) at presentation: a series of 158 cases from the Eurofever/EUROTRAPS international registry.	Ann Rheum Dis. 73(12):2160-7	2014	12,32	Gaslini Genova
Autoinflammatory syndromes	Piram M, Koné-Paut I, Lachmann HJ, Frenkel J, Ozen S, Kuemmerle-Deschner J, Stojanov S, Simon A, Finetti M, Sormani MP, Martini A, Gattorno M,	Validation of the auto-inflammatory diseases activity index (AIDAI) for hereditary recurrent fever syndromes	Ann Rheum Dis. 73(12):2168-73	2014	12,32	Gaslini Genova

	Ruperto N; EUROFEVER, EUROTRAPS and the Paediatric Rheumatology International Trials Organisation (PRINTO) networks					
Autoinflammatory syndromes	**Finetti M**, Insalaco A, Cantarini L, Meini A, Breda L, Alessio M, D'Alessandro M, Picco P, Martini A, **Gattorno M**	Long-term efficacy of interleukin-1 receptor antagonist (anakinra) in corticosteroid-dependent and colchicine-resistant recurrent pericarditis	J Pediatr. 164(6):1425-31.e1	2014	3,89	Gaslini Genova
Autoinflammatory syndromes	Federici S, **Gattorno M**	A practical approach to the diagnosis of autoinflammatory diseases in childhood.	Best Pract Res Clin Rheumatol. 28(2):263-76	2014	3,26	Gaslini Genova
Autoinflammatory syndromes	Hofer M, Pillet P, Cochard MM, Berg S, Krol P, Kone-Paut I, Rigante D, Hentgen V, Anton J, Brik R, Neven B, Touitou I, Kaiser D, Duquesne A, Wouters C, **Gattorno M**	International periodic fever, aphthous stomatitis, pharyngitis, cervical adenitis syndrome cohort: description of distinct phenotypes in 301 patients	Rheumatology (Oxford). 53(6):1125-9	2014	4,52	Gaslini Genova
Autoinflammatory syndromes	Omenetti A, Carta S, Delfino L, Martini A, **Gattorno M***, Rubartelli A*	Increased NLRP3-dependent interleukin 1β secretion in patients with familial Mediterranean fever: correlation with MEFV genotype	Ann Rheum Dis. ;73(2):462-9. (*last co-authors)	2014	12,32	Gaslini Genova
Autoinflammatory syndromes	Ozen S, Demirkaya E, Amaryan G, Koné-Paut I, Polat A, Woo P, Uziel Y, Modesto C, Finetti M, Quartier P, Papadopoulou-Alataki E, Al-Mayouf SM, Fabio G, Gallizzi R, Cantarini L, Frenkel J, Nielsen S, Hofer M, Insalaco A, Acikel C, Ozdogan H, Martini A, Ruperto N, **Gattorno M**; Paediatric Rheumatology International Trials Organisation; Eurofever Project	Results from a multicentre international registry of familial Mediterranean fever: impact of environment on the expression of a monogenic disease in children.	Ann Rheum Dis. 73(4):662-7	2014	12,32	Gaslini Genova

Autoinflammatory syndromes	Caorsi R, Picco P, Buoncompagni A, Martini A, **Gattorno M**	Osteolytic lesion in PAPA syndrome responding to anti-interleukin 1 treatment	J Rheumatol. 41(11):2333-4	2014	3,23	Gaslini Genova

Trial clinici farmacologici

***Trial* clinici del Gruppo di Studio di Reumatologia Pediatrica dal 2014 ad oggi**

È stata effettuata una revisione dei principali trial clinici in cui hanno partecipato o sono stati coordinatori componenti del Gruppo di studio (Gds) di Reumatologia Pediatrica.

La ricerca è stata ristretta ai trial di intervento farmacologico in atto nel periodo dal 01/01/2014 al 16/06/2017.

Sono stati quindi selezionati 19 trial clinici, 12 ancora aperti e 7 già conclusi: la principale area di ricerca è risultata essere l'artrite idiopatica giovanile, seguita in seconda istanza dalle patologie autoinfiammatorie e dalle vasculiti.

Tutti i trial selezionati sono multicentrici e di questi solo uno esclusivamente italiano. La maggior dei trial sono sponsorizzati (84%), tuttavia ve ne sono 3 di tipo accademico, senza sponsor di case farmaceutiche (AnakInRa for Treatment for Recurrent Idiopathic Pericarditis, AIRTRIP trial; Mycophenolate mofetil vs cyclophosphamide for polyarteritis nodosa, MYPAN trial; Anakinra for IvIg resistant children with Kawasaki disease, Kawakinra trial).

La maggior parte dei farmaci oggetto di studio sono farmaci biologici, con l'eccezione del micofenolato mofetile nello studio MYPAN. In particolare sono stati inclusi 5 trial su tocilizumab, 4 su canakinumab, 2 rispettivamente su anakinra e sarilumab e 1 trial per abatacept, adalimumab, rituximab, secukinumab e anticorpo monoclonate anti INFgamma.

Trial clinici farmacologici

1 Nome MR	2 *Sponsor*	3 *Partners*		4 Ruolo		5 Denominazione del *trial*	6 Farmaco	7 Durata		8 Note
		I	N	C	P			C	T (anno)	
Artrite idiopatica giovanile a decorso sistemico (sJIA)	Roche	x		x		Studio di 12 settimane, randomizzato, in doppio cieco, controllato placebo, a gruppi paralleli, a 2 bracci di trattamento per valutare l'efficacia e la sicurezza di tocilizumab in pazienti con artrite giovanile idiopatica sistemica (sJIA) attiva, con un periodo di estensione di 92 settimane, in aperto, a singolo braccio per esaminare l'utilizzo a lungo termine del tocilizumab. (WA18221)	Tocilizumab		2015	Gruppo di studio di reumatologia pediatrica
Artrite idiopatica giovanile a decorso sistemico (sJIA)	Roche	x		x		Studio di fase IV per valutare la riduzione della frequenza di somministrazione di Tocilizumab in pazienti affetti da Artrite Idiopatica Giovanile Sistemica (sJIA) che manifestano anomalie nei parametri di laboratorio durante il trattamento con Tocilizumab. (WA28029)	Tocilizumab	x		Gruppo di studio di reumatologia pediatrica
Granulomatosi con poliangite (Wegener's) (GPA) e poliangite microscopica (MPA)	Roche	x		x		Studio di fase IIa, per valutare la sicurezza e la farmacocinetica di 4 infusioni per via endovenosa di rituximab in pazienti pediatrici affetti da grave granulomatosi con poliangite (di Wegener) o poliangite microscopica. (WA25165)	Rituximab		2016	Gruppo di studio di reumatologia pediatrica

Artrite idiopatica giovanile a decorso poliarticolare (pJIA)	Roche	x		x		Studio di fase Ib, in aperto, multicentrico, per la valutazione della farmacocinetica, della farmacodinamica e della sicurezza di tocilizumab somministrato per via sottocutanea in pazienti affetti da artrite idiopatica giovanile a decorso poliarticolare. (WA28117)	Tocilizumab		2015	Gruppo di studio di reumatologia pediatrica
Pericardite ricorrente idiopatica	Maria Vittoria Hospital – Torino		x	x		AnakInRa for Treatment of Recurrent Idiopathic Pericarditis (AIRTRIP) Studio di 8 mesi, multicentrico (3 centri italiani), randomizzato, in doppio cieco, controllato placebo, per valutare l'efficacia, la tollerabilità e la sicurezza di anakinra in adulti e bambini con pericardite idiopatica ricorrente (RP) (2013-001849-13 EudraCT Number)	Anakinra		2015	Gruppo di studio di reumatologia pediatrica
Artrite idiopatica giovanile a decorso sistemico (sJIA)	Roche	x		X		Studio di fase Ib, in aperto, multicentrico, per la valutazione della farmacocinetica, della farmacodinamica e della sicurezza di Tocilizumab somministrato per via sottocutanea in pazienti affetti da Artrite Idiopatica Giovanile sistemica. (WA28118)	Tocilizumab	x		Gruppo di studio di reumatologia pediatrica
Artrite idiopatica giovanile a decorso sistemico (sJIA)/ Artrite idiopatica giovanile a decorso poliarticolare (pJIA)	Roche	x		x		Studio di estensione a lungo termine per valutare la sicurezza e l'efficacia di tocilizumab sottocute in pazienti con artrite idiopatica giovanile a decorso poliarticolare (pJIA) e sistemico (sJIA) (WA29231)	Tocilizumab	x		Gruppo di studio di reumatologia pediatrica

Artrite idiopatica giovanile a decorso sistemico (sJIA)	Novartis	x		x		Studio di estensione in aperto canakinumab (ACZ885) in pazienti con artrite idiopatica sistemica giovanile (SjIA) con manifestazioni sistemiche attive. (CACZ885G2301E1)	Canakinumab		2015	Gruppo di studio di reumatologia pediatrica
TNF receptor Associated Periodic Syndrome (TRAPS)	Novartis	x			x	Studio in aperto, multicentrico, per valutare l'efficacia e la sicurezza di un trattamento con Canakinumab per 4 mesi con follow-up di 5 mesi in pazienti con sindrome periodica attiva ricorrente o cronica associata al recettore del TNF (TRAPS) - (CACZ885D2203)	Canakinumab		2014	Gruppo di studio di reumatologia pediatrica
Febbri periodiche (TRAPS, HIDS, crFMF)	Novartis	x			x	Studio randomizzato, in doppio cieco, controllato verso placebo con canakinumab in pazienti con febbri periodiche ereditarie (TRAPS, HIDS oppure crFMF), con successivo periodo randomizzato di sospensione/riduzione della frequenza di somministrazione del trattamento, e periodo di trattamento a lungo termine in aperto (CACZ885N2301)	Canakinumab	x		Gruppo di studio di reumatologia pediatrica
Artrite idiopatica giovanile a decorso sistemico (sJIA)/	Novartis	x		x		Studio in aperto di efficacia e sicurezza sulla riduzione della dose o il prolungamento dell'intervallo di somministrazione di canakinumab (ACZ885) in pazienti con artrite idiopatica giovanile sistemica (SJIA) - (CACZ885G2306)	Canakinumab	x		End of Study expected by September 2017

Artrite idiopatica giovanile associate a entesite (ERA) / Artrite psoriasica (PsA)	Novartis	x		x		Studio in tre parti randomizzato, in doppio cieco, controllato verso placebo, per indagare l'efficacia e la sicurezza della terapia con secukinumab per due tipi di artrite idiopatica giovanile: artrite psoriasica e artrite entesite-relata (CAIN457F2304)	Secukinumab	x		Gruppo di studio di reumatologia pediatrica
Artrite idiopatica giovanile a decorso poliarticolare (pJIA)	Abbott	x		x		Registro a lungo termine di HUMIRA (Adalimumab) nell'artrite idiopatica giovanile (AIG): studio osservazionale, non interventistico, di coorte prospettico, multicentrico, per valutare la sicurezza e l'efficacia a lungo termine di HUMIRA® (Adalimumab) in pazienti con artrite idiopatica giovanile (AIG) poliarticolare attiva da moderata a severa o con AIG con decorso poliarticolare (P10-262)	Adalimumab	x		Gruppo di studio di reumatologia pediatrica
Artrite idiopatica giovanile a decorso poliarticolare (pJIA)	Bristol-Myers Squibb	x		x		Studio di fase 3, multicentrico, in aperto per valutare la farmacocinetica, l'efficacia e la sicurezza di abatacept sottocute (SC) in bambini e adolescenti con artrite idiopatica giovanile a decorso poliarticolare in fase attiva (pJIA) e risposta insufficiente a farmaci biologici o non biologici (DMARDs) (NCT01844518)	Abatacept		2016	Gruppo di studio di reumatologia pediatrica
Artrite idiopatica giovanile a decorso poliarticolare (pJIA)	Sanofi	x			x	Studio in aperto, sequenziale, a dose crescente per identificare la dose terapeutica per il Sarilumab per via sottocutanea in bambini e adolescenti, tra i 2 e 17 anni, con artrite idiopatica giovanile a decorso poliarticolare (pJIA), seguito da uno studio di estensione (DRI13925)	Sarilumab	x		Gruppo di studio di reumatologia pediatrica

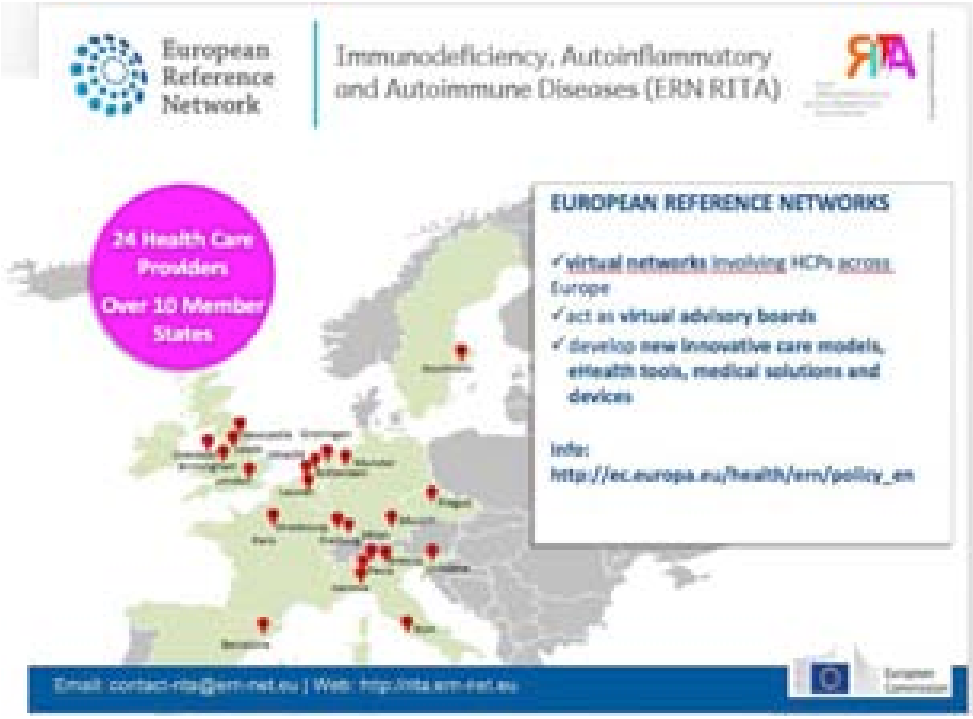
Artrite idiopatica giovanile a decorso sistemico (sJIA)	Sanofi	x			x	Studio in aperto, sequenziale, a dose crescente per identificare la dose terapeutica per il Sarilumab per via sottocutanea in bambini e adolescenti, tra i 2 e 17 anni, con artrite idiopatica giovanile a decorso sistemico (sJIA), seguito da uno studio di estensione (DRI13926)	Sarilumab	x		Gruppo di studio di reumatologia pediatrica
Panarterite nodosa giovanile (PAN)	University of London		x		x	Studio clinico controllato e randomizzato in aperto di micofenolato mofetile (MMF) contro ciclofosfamide (CYC) per l'induzione della remissione della poliarterite nodosa giovanile (PAN) ISRCTN75434563	Micofenolato mofetile	x		Gruppo di studio di reumatologia pediatrica
Malattia di Kawasaki	Hopitaux de Paris		X		x	Sudio di fase 2, multicentrico per valutare l'efficacia e la sicurezza di anakinra nei pazienti con malattia di Kawasaki resistenti al trattamento con infusione di immunoglobuline (AOM13520)	Anakinra	x		Gruppo di studio di reumatologia pediatrica
Sindrome da attivazione macrofagica (MAS) / linfoistiocitosi emofagocitica secondaria (sHLH)	Novimmune	x		x		Studio pilota, in aperto, singolo braccio di studio, multicentrico, per valutare la sicurezza, la tollerabilità, la farmacocinetica e l'efficacia dell'infusione per via endovenosa di NI-0501, un anticorpo monoclonale anti-interferon gamma (anti-IFNγ), in pazienti con syndrome da attivazione macrofagica (MAS) in corso di artrite idiopatica giovanile sistemica (sJIA) /HLH secondaria (sHLH)	Anti-INFγ Abm	x		Gruppo di studio di reumatologia pediatrica

Partecipazione Nazionale a ERN

Le Sindromi Autoinfiammatorie

Marco Gattorno
U.O Pediatria II dell'Ospedale "Gaslini" e Dipartimento di Pediatria dell'Università di Genova

Con questo termine si definiscono alcune affezioni, ad ereditarietà monogenica, caratterizzate da una alterazione dei meccanismi di controllo della risposta infiammatoria. Tali malattie possono assumere un andamento periodico o ricorrente (Febbri periodiche), caratterizzato da accessi febbrili spesso accompagnati da sintomatologia muco-cutanea, gastrointestinale e articolare (Febbre Familiare Mediterranea, Sindrome TRAPS, Sindrome da IperIgD). In altre situazioni la componente infiammatoria invece ad assumere un carattere cronico e sistemico, con coinvolgimento di diversi organi o apparati (S. di Muckle-Wells o Sindrome CINCA). In un terzo gruppo di malattie, la componeNte infiammatoria tende ad essere prevalentemente localizzata a specifiche strutture, quali le articolazioni, la cute o l'occhio (sindromi di Blau, PAPA, DIRA, DITRA, Majeed). Recentemente sono state evidenziate entità cliniche monogeniche associate a interessamento vascolare, come il difetto di ADA2, legato ad una poliarterite ad esordio precoce associata a stroke e le cosidette Interferonopate (sindrome di Candle e SAVI), secondarie ad una disregolazione dei meccanismi di attivazione dell'interferone di tipo I.
Dal 2009 esiste un registro internazionale, Eurofever (www.printo.it/eurofever). Il registro raccoglie 3650 pazienti pediatrici ed adulti da 108 centri in 39 paesi europei ed extra-europei. I dati raccolti hanno permesso la pubblicazione delle più ampie casistiche di malattie autoinfiammatorie monogeniche, di riportare le risposte ai trattamenti biologici e non in uso in queste patologia e la messa a punto di nuovi criteri classificativi *evidence-based*. Tale registro rappresenta l'ideale strumento per incrementare una collaborazione sempre più stretta tra le diverse patologie rare di natura immunologica, nell'ambito dell'European Reference Network (ERN).


European Reference Network
Immunodeficiency, Autoinflammatory and Autoimmune Diseases (ERN RITA)
24 Health Care Providers
Over 10 Member States
EUROPEAN REFERENCE NETWORKS
✓virtual networks involving HCPs across Europe
✓act as virtual advisory boards
✓develop new innovative care models, eHealth tools, medical solutions and devices
info:
http://ec.europa.eu/health/ern/policy_en
Email: contact-rita@ern-net.eu | Web: http://rita.ern-net.eu


ERN-RITA
24 Health Care Providers
Over 10 Member States

Malattie cardiologiche pediatriche

A cura della Società Italiana di Cardiologia Pediatrica – SICP
(Presidente: *Maria Giovanna Russo*)

Giuseppe Limongelli
M. Giovanna Russo

Malattie Cardiovascolari- Pubblicazioni

Società Italiana
di Cardiologia Pediatrica
e delle Cardiopatie Congenite

Produzione Scientifica Gennaio 2016 – Maggio 2017

Nel periodo di tempo considerato i Soci della SICP hanno prodotto **94 Lavori Scientifici. Per 75 di questi è stato possibile calcolare l'IF ottenendo un IF totale di: 342,446 con una media di 4,56 + 10,1 (range 0,811 – 59,558).**
Il valore più elevato di IF è stato ottenuto da due articoli pubblicati sul NEJM, entrambi nel 2017, mentre per il 2016 l'articolo che ha ottenuto l'IF più elevato è stato pubblicato su JAMA.
30 articoli sono stati pubblicati su Riviste con IF > 3 (40%)
45 articoli sono stati pubblicati su Riviste con IF > 2 (60%).

Società Italiana
di Cardiologia Pediatrica
e delle Cardiopatie Congenite

Paragonando il 2016 con i primi 6 mesi del 2017 si nota immediatamente la veloce progressione della Società e l'incremento di importanza degli articoli.
Infatti nel **2016 l'IF totale è stato di 124,23 con una media di 3,882 per articolo; mentre nei primi 6 mesi del 2017 l'IF totale è già di 218,216 con una media per articolo di 5,07.**
La produzione scientifica del 2017 rispetto a quella del 2016 appare molto più elevata persino se non si valutano i due articoli del NEJM ottenendo un IF totale di 100,1 che, seguendo il trend, porterebbe per la fine dell'anno in corso ad un eccellente **IF totale di 201 con un aumento percentuale rispetto allo scorso anno di circa +70%.**

Malattie Cardiovascolari - Partecipazione Nazionale a ERN

Giuseppe Limongelli
AORN Monaldi – Università della Campania Luigi Vanvitelli

Maria Giovanna Russo
AORN Monaldi – Università della Campania Luigi Vanvitelli - Presidente SICP

Le malattie rare del cuore rappresentano un settore per tanti anni "orfano" dell'interesse dei cardiologi pediatri e dell'adulto, ma che ad oggi sta iniziando ad avere sempre maggiore attenzione.
Cardiopatie congenite, cardiomiopatie, aortopatie, malattie del pericardio, malattie dei canali ionici, malattie ereditarie associate a morte improvvisa cardiaca giovanile, sono tutte condizioni rare, ma che spesso sono gravate da alto tasso di "morbilità" (necessità di intervento, complicanze) e mortalità (morte improvvisa, morte peri-post operatoria, scompenso cardiaco).
In Italia, l'*expertise* è elevatissimo nelle cardiopatie congenite, nelle cardiomiopatie pediatriche, nelle malattie dei canali ionici. Non c'è grossa esperienza *in fieri* di *trial* pediatrici, ma all'orizzonte si affacciano nuove prospettive nel campo delle cardiomiopatie e dei canali ionici. Anche le malattie cardiache hanno una loro rete europea.
La rete ERN per le malattie cardiache è stata denominata GUARD-HEART (Gateway to Uncommon And Rare Diseases of the **HEART**) ed è coordinata dal Dr. Arthur Wilde presso l'Academic Medical Centre di Amsterdam.
Le malattie sono state classificate in 4 sottocategorie che comprendono:

1. Cardiomiopatie Adulti
2. Aritmie (Disturbi Elettrici) Familiari
3. Patologie pediatriche

Considerando che la il DL 279/2001, non comprende tra le malattie del sistema circolatorio nessuno di queste condizioni, per aderire alla rete, ed esser inquadrati in una o piu' categorie, i centri hanno dovuto certificare (attraverso la presentazione di dati, SDO, partecipazioni e registri riconosciuti, etc) di seguire un numero target di pazienti, di avere una produzione scientifica e/o un ruolo di leader in studi clinici inerenti alla specifica categoria. Tra i 24 centri europei complessivi, in Italia, i centri pediatrici che afferiscono al momento alla rete sono: Padova (Clinica Pediatrica – Azienda Ospedaliera di Padova), Roma (Ospedale Pediatrico Bambino Gesù), e Napoli (Ospedale Monaldi/ AORN Colli/Università della Campania). Il primo obiettivo è stato raggiunto con la creazione di un "Registro sul QT lungo" proposto dalla Dr.ssa Sequella Brugada all'ultimo incontro di Vilnius.

http://guardheart.ern-net.eu/special-electrophysiology-conditions/

NETWORK APPLICATION FORM

5. List all the Healthcare Providers applying for membership in the Network Applicant and their location.

N°	Name of Healthcare Provider	Name of the Healthcare Provider Representative	Category of Facility, i.e. Regional Hospital, University Hospital, Private Clinic, etc.	Member State
1	Academic Medical Center	Arthur Wilde	University Hospital	Netherlands
2	Sant Joan de Déu - Barcelona Children's Hospital	Georgia Sarquella-Brugada	University Hospital	Spain
3	IRCCS Fondazione Salvatore Maugeri	Silvia Priori	University Hospital	Italy
4	Assistance Publique Hôpitaux de Paris	Philippe Charron	University Hospital	France
5	Hospital Universitario Puerta de Hierro Majadahonda	Pablo Garcia	University Hospital	Spain
6	CHU de Nantes	Vincent Probst	University Hospital	France
7	The Hospital District of Helsinki and Uusimaa	Jaana Pihkala	University Hospital	Finland
8	Motol University Hospital	Jan Janousek	University Hospital	Czech Republic
9	University Hospitals Leuven	Rik Willems	University Hospital	Belgium
10	Ospedale Pediatrico Bambino Gesù	Fabrizio Drago	University Hospital	Italy
11	Umea University Hospital	Annika Rydberg	University Hospital	Sweden
12	Copenhagen University Hospital	Jacob Tfelt	University Hospital	Denmark
13	University Hospital Virgen Arrixaca	Juan Ramon Gimeno	University Hospital	Spain
14	University Hospital Munster	Eric Schulze-Bahr	University Hospital	Germany
15	St George's University Hospitals NHS Foundation Trust	Elijah Behr	University Hospital	United Kingdom
16	HCL hospital cardiologique CHU de Lyon	Philippe Chevalier	University Hospital	France
17	Emergency Institute for cardiovascular disease: Prof.dr.C.C.Iliescu	Ruxandra Jurcut	University Hospital	Romania
18	Azienda Ospedaliera di Rilievo Nazionale (AORN) dei colli - Ospedale Monaldi	Guiseppe Limongelli	University Hospital	Italy