

PERSONAL INFORMATION

Federica Ricci

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Sex Female | Date of birth 04/06/1978 | Nationality Italian

WORK EXPERIENCE

- 12/31/2010-today **Medical Director as Child Neuropsychiatrist**
University Hospital "Città della Salute e della Scienza di Torino", Children Hospital "Regina Margherita", Department of Public Health Sciences and Pediatrics, Piazza Polonia 94 10126 Torino (Italy)
Field: neuromuscular disorder (responsible for the infant sector of the neuromuscular diseases center), acquired brain injury, neurophysiology.
- 2017-today **Lecturer for the Medical Specialization Course in Child Neurology and Psychiatry**
University of Turin, discipline "Neurology of first and second childhood" (total 7 hours)
- 2015-today **Lecturer for the Degree Course in Physiotherapy**
University of Turin, discipline "Child Neuropsychiatry" (total 12 hours)
- 2012-today **Lecturer for the Degree Course in Developmental Neuro and Psychomotor Therapy**
University of Turin, "Rehabilitation of neuromuscular diseases and special kinesiology" (total 20 hours)
- 04/01/2009-12/30/2010 **Lecturer for the Degree Course in Audiometric Techniques and Hearing Aid Techniques**
University of Turin, discipline "Child Neuropsychiatry" (total 12 hours)
- 04/01/2004-12/30/2010 **Expert consultant at the Rehabilitation Visual Center**
Civil Hospital of Ivrea (Turin, Italy), as a medical specialist in Child Neuropsychiatry, for a total of 50 hours per month
- 2004-2009 **General Medical Practitioner for Continuity of Health Care activities**
Health district of Ivrea (Turin, Italy)
- 2012-2014 **Voluntary activities in Gulu, Northern Uganda, for the project "Rehabilitation in Northern Uganda to improve the quality of treatment of disabled and epileptic patients in the hospital and community rehabilitation (CBR)"**
4 short missions (December 2004-January 2005, August 2005, September 2007, April-May 2009) organized by the NGO "Comitato Collaborazione Medica" (Turin, Italy)
- Expert consultant free of charge in Gulu, Northern Uganda for diagnosis and setting for the treatment of epileptic children and disabled at health facilities in the Gulu District**
2 short missions (from 11/29/2012 to 12/14/2012 and from 11/23/2014 to 07/12/2014) for the NGO "Comitato Collaborazione Medica" (Turin, Italy)

EDUCATION AND TRAINING

- 03/16/2013 **Master's Degree in "Child Rehabilitation and research methodology", University of Modena and Reggio Emilia**
Score: 110/110 magna cum laude

- 12/17/2008 Specialist Degree in Child Neuropsychiatry, University of Turin, Faculty of Medicine
Score: 70/70 magna cum laude and dignity of the press
Thesis title: "SMARD1: a new and rare disease entity in the field of neuromuscular disease"
- 12/19/2003 Licence to practice, Turin, Italy (Licence number: 20108)
- 07/21/2003 Degree in Medicine and Surgery, University of Turin, Faculty of Medicine
Score: 110/110 magna cum laude and dignity of the press
Thesis title: "Congenital cerebellar pathology: etiopathogenetic, nosographic and clinical issues"
- July 1997 Scientific High School Diploma, "A.Gramsci" High School, Ivrea (Turin, Italy)
Score: 60/60

PERSONAL SKILLS

Mother tongue Italian

Other language(s)

	UNDERSTANDING		SPEAKING		WRITING
	Listening	Reading	Spoken interaction	Spoken production	
English	B2	B2	B2	B2	B2

Levels: A1/A2 basic users, B1/B2 independent users, C1/C2 proficient users

ADDITIONAL INFORMATION

Research activity

Principal Investigator in research project "Trial readiness and endpoint assessment in congenital and childhood Myotonic Dystrophy" supported by Telethon and coordinated by Dr. V.Sansone (GUP 19002H, NEMO center, Milan)

Principal Investigator in multicentre phase IV study "Long-Term Observational Study of Translarna Safety and Effectiveness in Usual Care" supported by PTC Therapeutics and coordinated by Prof. E.Mercuri (Pediatric Hospital Bambino Gesù, Rome)

Principal Investigator in multicenter observational study "Long term natural history in Duchenne muscular dystrophy" supported by Telethon and coordinated by Prof. E.Mercuri (GUP 15011N, Pediatric Hospital Bambino Gesù, Rome)

Principal Investigator in multicenter research project "Complete molecular characterization of patients affected by congenital muscular dystrophies with alphadystroglycan defect using next generation sequencing strategies" supported by Telethon and coordinated by Dr. A.D'Amico (GUP 13004L, Pediatric Hospital Bambino Gesù, Rome)

Trial Coordinator in multicenter clinical trial "FOR DMD, Duchenne muscular dystrophy: double-blind randomized controlled trial to find optimum steroid regimen" supported by United States National Institutes of Health, National Institute of Neurological Disorders and Stroke (NINDS) and coordinated by Prof. K. Bushby (Newcastle upon Tyne, UK) and Prof. RC Griggs (Rochester, NY)

Clinical Evaluator in multicenter research project "Development of a registry and a database for a nation-wide Italian collaborative network on congenital muscular dystrophy" supported by Telethon and coordinated by Prof.E.Mercuri (GUP11001F, Policlinico Gemelli, Rome)

Board member and consultant of the Rehabilitation in Pediatric Onco-hematology Work Group of the Italian Association of Pediatric Oncology and Hematology (AIEOP)

Member of the LICE (Italian League Against Epilepsy) Study Group "Epilepsy and brain tumors" coordinated by Dr. M.Maschio and Prof. F.Paladin, Rome

Publications

Intrathecal administration of nusinersen in adult and adolescent patients with spinal muscular atrophy and scoliosis: Transforaminal versus conventional approach. *Neuromuscul Disord.* 2019 Oct;29(10):742-746. Bortolani S, Stura G, Ventili G, Vercelli L, Rolle E, **Ricci F**, Bergui M, Mongini T.

A new frontier of pediatric oncology rehabilitation: Establishment of a dedicated working-group within the Italian Association of Pediatric Hematology and Oncology. *J Pediatr Rehabil Med.* 2019;12(3):271-277. Rossi F, Coppo M, Zucchetti G, **Ricci F**, Fagioli F.

Long-term natural history data in Duchenne muscular dystrophy ambulant patients with mutations amenable to skip exons 44, 45, 51 and 53. *PLoS One.* 2019 Jun 25;14(6):e0218683. Brogna C, Coratti G, Pane M, Ricotti V, Messina S, D'Amico A, Bruno C, Vita G, Berardinelli A, Mazzone E, Magri F, **Ricci F**, Mongini T, Battini R, Bello L, Pegoraro E, Baranello G, Previtali SC, Politano L, Comi GP, Sansone VA, Donati A, Bertini E, Muntoni F, Goemans N, Mercuri E; on behalf on the International DMD group.

New pharmacotherapies for genetic neuromuscular disorders: opportunities and challenges. *Expert Rev Clin Pharmacol.* 2019 Jun 21. doi: 10.1080/17512433.2019.1634543. [Epub ahead of print]. **Ricci F**, Vacchetti M, Brusa C, Vercelli L, Davico C, Vitiello B, Mongini T.

Molecular Genetics and Interferon Signature in the Italian Aicardi Goutières Syndrome Cohort: Report of 12 New Cases and Literature Review. *J Clin Med.* 2019 May 26;8(5). Garau J, Cavallera V, Valente M, Tonduti D, Sproviero D, Zucca S, Battaglia D, Battini R, Bertini E, Cappanera S, Chiapparini L, Crasà C, Cricchiutti G, Giustina ED, D'Arrigo S, Giorgis V, Simone M, Galli J, Piana R, Messina T, Moroni I, Nardocci N, Panteghini C, Parazzini C, Pichiecchio A, Pini A, **Ricci F**, Saletti V, Salvatici E, Santorelli FM, Sartori S, Tinelli F, Uggetti C, Veneselli E, Zorzi G, Garavaglia B, Fazzi E, Orcesi S, Cereda C

Early higher dosage of alglucosidase alpha in classic Pompe disease. *J Pediatr Endocrinol Metab.* 2018 Nov 15. doi: 10.1515/jpem-2018-0336. [Epub ahead of print] Spada M, Pagliardini V, **Ricci F**, Biamino E, Mongini T, Porta F.

A method for intraoperative recording of the laryngeal adductor reflex during lower brainstem surgery in children. *Clin Neurophysiol.* 2018 Sep 24. doi: 10.1016/j.clinph.2018.08.028. [Epub ahead of print] Costa P, Gaglini PP, Tavormina P, **Ricci F**, Peretta P.

Functional assessment tools in children with Pompe disease: A pilot comparative study to identify suitable outcome measures for the standard of care. *Eur J Paediatr Neurol.* 2018 Aug 18. doi: 10.1016/j.ejpn.2018.08.001. [Epub ahead of print] **Ricci F**, Brusa C, Rossi F, Rolle E, Placentino V, Berardinelli A, Pagliardini V, Porta F, Spada M, Mongini T.

Upper limb function in Duchenne muscular dystrophy: 24 month longitudinal data. *PLoS One.* 2018 Jun 20;13(6). Pane M, Coratti G, Brogna C, Mazzone ES, Mayhew A, Fanelli L, Messina S, D'Amico A, Catteruccia M, Scutifero M, Frosini S, Lanzillotta V, Colia G, Cavallaro F, Rolle E, De Sanctis R, Forcina N, Petillo R, Barp A, Gardani A, Pini A, Monaco G, D'Angelo MG, Zanin R, Vita GL, Bruno C, Mongini T, **Ricci F**, Pegoraro E, Bello L, Berardinelli A, Battini R, Sansone V, Albamonte E, Baranello G, Bertini E, Politano L, Sormani MP, Mercuri E.

Differential diagnosis of vacuolar muscle biopsies: use of p62, LC3 and LAMP2 immunohistochemistry. *Acta Myol.* 2017 Dec 1;36(4):191-198. Vittonatto E, Boschi S, Chiadò-Piat L, Ponzalino V, Bortolani S, Brusa C, Rainero I, **Ricci F**, Vercelli L, Mongini T.

Patterns of care of brain tumor-related epilepsy. A cohort study done in Italian Epilepsy Center. *PLoS One* 12(7), 17 luglio 2017. Maschio M, Beghi E, Casazza MML, Colicchio G, Costa C, Banfi P, Quadri S, Aloisi P, Giallonardo AT, Buttinelli C, Pauletto G, Striano S, Salmaggi A, Terenzi R, Daniele O, Cricchiutti G, Paladin F, Rossi R, Prato G, Vigevano F, De Simone R, **Ricci F**, Saladini M, Monti F, Casellato S, Zanoni G, Giannarelli D, Avanzini G, Aguglia U; BTRE Study Group

Diagnosis of Duchenne Muscular Dystrophy in Italy in the last decade: Critical issues and area of improvements.

Neuromuscular Disorders 27, 447-451, 2017. D'Amico A, Catteruccia M, Baranello G, Politano L, Govoni A, Previtali SC, Pane M, D'Angelo MG, Bruno C, Messina S, **Ricci F**, Pegoraro E, Pini A, Berardinelli A, Gorni K, Battini R, Vita G, Trucco F, Scutifero M, Petillo R, D'Ambrosio P, Ardisson A, Pasanisi B, Vita G, Mongini T, Moggio M, Comi GP, Mercuri E, Bertini E

Registries versus tertiary care centers: How do we measure standards of care in Duchenne muscular dystrophy?

Neuromuscular Disorders 26, 261-263, 2016. Mercuri E, Baranello G, Battini, Berardinelli A, Bertini E, Bruno C, Comi GP, D'Amico A, D'Angelo G, Gorni K, Messina S, Mongini T, Pane M, Pegoraro E, Pini A, Politano L, Previtali S, **Ricci F**, Sansone V, Vita GL

Impaired Presynaptic High-Affinity Choline Transporter Causes a Congenital Myasthenic Syndrome with Episodic Apnea.

The American Journal of Human Genetics 99, 1-9, September 1, 2016. Bauché S, O'Regan S, Azuma Y, Laffargue F, McMacken G, Sternberg D, Brochier G, Buon C, Bouzidi N, Topf A, Lacène E, Remerand G, Beaufre AM, Pebrel-Richard C, Thevenon J, El Chehadeh-Djebbar S, Faivre L, Duffourd Y, **Ricci F**, Mongini T, Fiorillo C, Astrea G, Burloiu CM, Butoianu N, Sandu C, Servais L, Bonne G, Nelson I, Desguerre I, Nougues MC, Bœuf B, Romero N, Laporte J, Boland A, Lechner D, Deleuze JF, Fontaine B, Strohlic L, Lochmuller H, Eymard B, Mayer M, Nicole S.

Rehabilitative intervention during and after pediatric hematopoietic stem cell transplantation: an analysis of the existing literature.

Pediatric Blood and Cancer, 2016. F.Rossi, M.Coppo, G.Zucchetti, D.Bazzano, **F.Ricci**, E.Vassallo, F.Nesi, F.Fagioli

Taking care of patients with brain tumor-related epilepsy: results from an Italian survey. Neurological Sciences, January 2015, Volume 36, Issue 1, pp 125-130. Maschio M, Paladin F, on behalf of the LICE Brain tumor-related epilepsy study group.

Characterization of human disease phenotypes associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR, and IFIH1.

American Journal of Medical Genetics, 2015 Feb;167A(2):296-312. Crow YJ, Chase DS, Lowenstein Schmidt J, Szynkiewicz M, Forte GM, Gornall HL, Oojageer A, Anderson B, Pizzino A, Helman G, Abdel-Hamid MS, Abdel-Salam GM, Ackroyd S, Aeby A, Agosta G, Albin C, Allon-Shalev S, Arellano M, Ariaudo G, Aswani V, Babul-Hirji R, Baildam EM, Bahi-Buisson N, Bailey KM, Barnerias C, Barth M, Battini R, Beresford MW, Bernard G, Bianchi M, Billette de Villemeur T, Blair EM, Bloom M, Burlina AB, Carpanelli ML, Carvalho DR, Castro-Gago M, Cavallini A, Cereda C, Chandler KE, Chitayat DA, Collins AE, Sierra Corcoles C, Cordeiro NJ, Crichtutti G, Dabydeen L, Dale RC, D'Arrigo S, De Goede CG, De Laet C, De Waele LM, Denzler I, Desguerre I, Devriendt K, Di Rocco M, Fahey MC, Fazzi E, Ferrie CD, Figueiredo A, Gener B, Goizet C, Gowrinathan NR, Gowrishankar K, Hanrahan D, Isidor B, Kara B, Khan N, King MD, Kirk EP, Kumar R, Lagae L, Landrieu P, Lauffer H, Laugel V, La Piana R, Lim MJ, Lin JP, Linnankivi T, Mackay MT, Marom DR, Marques Lourenço C, McKee SA, Moroni I, Morton JE, Moutard ML, Murray K, Nabbout R, Nampoothiri S, Nunez-Enamorado N, Oades PJ, Olivier I, Ostergaard JR, Pérez-Dueñas B, Prendiville JS, Ramesh V, Rasmussen M, Régal L, **Ricci F**, Rio M, Rodriguez D, Roubertie A, Salvatici E, Segers KA, Sinha GP, Soler D, Spiegel R, Stöberg TI, Straussberg R, Swoboda KJ, Suri M, Tacke U, Tan TY, te Water Naude J, Wee Teik K, Thomas MM, Till M, Tonduti D, Valente EM, Van Coster RN, van der Knaap MS, Vassallo G, Vijzelaar R, Vogt J, Wallace GB, Wassmer E, Webb HJ, Whitehouse WP, Whitney RN, Zaki MS, Zuberi SM, Livingston JH, Rozenberg F, Lebon P, Vanderver A, Orcesi S, Rice GI

Cerebral Palsy in the Developing World. M.T.Perenchio and **F.Ricci** in "Orthopedic Management of Children with Cerebral Palsy. A comprehensive approach", Federico Canavese and Jacques Deslandes, Nova Science Publishers, 2015

Scientific conferences/invited speaker

Annual Congress Macroarea Nord-Ovest SINP organized by SINP, Cuneo, 10/29/2019

XIX National Congress AIM (Italian Association of Myology) organized by AIM, Bergamo, 06/05-08/2019

SMAcademy -Spinal Muscular Atrophy: new phenotypes and new implications organized by Edra Spa, Rome, 03/21-22/2019

Regional Congress of Italian Society of Neuropsychiatry of Childhood and Adolescence, organized by SINPIA, Turin, 02/15/2019

Regional Congress of Italian Society of Pediatrics, organized by SIP, Turin, 17/11/2018

Training in pediatrics, organized by Città della Salute e della Scienza di Torino, FIMP, SIP, Turin 24/10/2018

Pediatric traumatic brain injury, organized by SIMEUP, Turin, 06/06/2018

Neuromuscular Diseases Day, organized by Neuromuscular Alliance, AISNP, AIM and Telethon, Turin, 03/10/2018

The improvement of the clinical and therapeutic approach for neuromuscular diseases: the turning point of the new millennium, organized by UILDM, Turin, 10/28/2017

Master's degree course in Pediatric Emergencies 2016-2017, organized by the University of Turin, Turin, 05/04/2017

Neuromuscular Diseases Day, organized by Neuromuscular Alliance, AISNP, AIM and Telethon, Turin, 03/04/2017

The rehabilitation of acquired brain injuries in the developmental age: a project for holistic intervention, organized by Regina Margherita Children Hospital, Health District City of Turin and Puzzle Cooperative ONLUS, Turin, 03/03/2017

Immune pathology and orphan diseases 2017, organized by CMID, Turin, 01/25-28/2017

Starting from a correct neurological examination: neuromuscular diseases, organized by CPG srl, Turin, 11/02/2016

3rd National Rehabilitation Network Meeting in Pediatric Oncohematology, organized by the University Hospital "Città della Salute e della Scienza di Torino", Turin, 10/28/2016

News and views. Neuromuscular pathology from the child to adulthood: new tools and new opportunities, organized by the IRCCS Fondazione Stella Maris, Pisa, 09/23-24/2016

Importance of early diagnosis in neuromuscular diseases with new therapeutic approaches: 'red flags' for pediatricians and family doctors, organized by the Rare Diseases Commission of the Turin OMCeO, Turin, 07/08/2016

Decennial of the Master in Child Rehabilitation and Research Methodology, organized by the University of Modena and Reggio Emilia, Reggio Emilia, 05/19-20/2016

Rehabilitation Network Project in Pediatric Onco-haematology, organized by the University Hospital "Città della Salute e della Scienza di Torino", Turin, 10/30/2015

Territorial Meeting on Muscular Dystrophy of Duchenne and Becker, organized by Duchenne Parent Project, Genoa, 09/19/2015

University Master Degree Course on Rare Diseases, organized by the University of Turin, 02/27/2015

2nd module of the Global Health Course 2014, organized by the NGOs Health Board Consortium Piedmont, Turin, 06/20/2014

Training Course Diagnosis and assistance on rare diseases for pediatricians and general practitioners, organized by the Italian Federation of Rare Diseases, Turin, 10/10/2013

The pediatric caregiver: an excellence of the Regina Margherita Children's Hospital, organized by the University Hospital "Città della Salute e della Scienza di Torino", Turin, 05/15/2013

3rd module of the course of Tropical Medicine 2012, organized by the NGOs Health Board Consortium Piedmont, Turin, 10/17/2012

And now I'm going home ... the continuity of care in the complex pediatric pathology, organized by the University Hospital "Città della Salute e della Scienza di Torino", Turin, 10/24/2012, 11/07/2012, 12/19/2012

Pathways of treatment in neuromuscular diseases: the health network of the Piedmont region, organized by Faculty of Medicine and Surgery of Turin, Turin, 04/04/2012

Theoretical-Practical Course on intraoperative neurophysiological monitoring in neurosurgery (Milan, Italy, 03/14-16/2019)

18th National Congress of Italian Association of Myology (Genova, Italy, 06/06/2018-06/09/2018)

SMAcademy: from diagnosis to treatment (Milan, Italy, 03/22-23/2018)

Sarepta European Advisory Board (Rome, Italy, 03/21-22/2018)

EvoCas9: gene therapy made in Trento e perspectives for CMT (Trento, Italy, 03/17/2018)

Intraoperative Neurophysiology in Neurosurgery: Innovation, Controversies, and Future Perspectives (Verona, Italy, 12/14-16/2017)

8th Theoretical-Practical Course on Neurophysiological Monitoring in Intensive Care (Bergamo, Italy, 11/16-18/2017)

DMD Expert Meeting PTC (Rome, Italy, 06/09/2017)

SMAcademy: from diagnosis to treatment (Rome, Italy, 09/24-25/2017)

FOR-DMD study investigator meeting (Newcastle Upon Tyne, UK, 06/6-7/2017)

17th National Congress of Italian Association of Myology (Siracusa, Italy, 05/31/2017-06/03/2017)

Pompe disease - what has changed after 10 years of Replacement Enzymatic Replacement Therapy (Parma, Italy, 04/5-6/2017)

XIX Telethon Scientific Convention (Riva del Garda, Italy, 03/13-15/2017)

VIII Theoretical-Practical Course on intraoperative neurophysiological monitoring in neurosurgery (Mestre, Italy, 12/12-14/2016)

DMD Expert Meeting PTC (Rome, Italy, 10/18/2016)

Expert Masterclass on Duchenne Muscular Dystrophy (Warsaw, Poland, 09/8-9/2016)

16th National Congress of Italian Association of Myology (Lecce, Italy, 06/08-11/2016)

3rd International Residential Course on Drug Resistant Epilepsies (Tagliacozzo, Italy, 05/8-14/2016)

15th National Congress of Italian Association of Myology (Naples, Italy, 05/20-23/2015)

XVIII Telethon Scientific Convention (Riva del Garda, Italy, 03/9-11/2015)

14th National Congress of Italian Association of Myology (Sirmione, Italy, 05/08-10/2014)

13th National Congress of Italian Association of Myology (Stresa, Italy, 05/16-18/2013)

XVII Telethon Scientific Convention (Riva del Garda, Italy, 03/11-13/2013)

22nd Basic Course in Electroencephalography and Clinical Epileptology (Gargnano, Italy, 11/6-11/2011)

Memberships

Italian Society of Neuropsychiatry of Childhood and Adolescence (SINPIA) 2019
Italian Society of Pediatric Neurology (SINP) 2019
Italian Association of Myology (AIM) 2019
Italian League Against Epilepsy (LICE) 2019
Italian Society of Clinical Neurophysiology (SINC) 2019
Italian Association Pediatric Oncology and Hematology (AIEOP) 2019

