EUROPEAN CURRICULUM VITAE FORMAT

PERSONAL INFORMATION

Name	**SCALA MARCELLO**
Address	
Telephone	
Fax	
E-mail	
Nationality	Italian
Place and date of birth	NAPLES, 19 OCTOBER 1987

WORK EXPERIENCE

• Dates (from – to)	**2019-present**
• Name and address of employer	Università degli Studi di Genova
• Type of business or sector	PhD: neuromuscular, neurometabolic, and neurodegenerative disorders in pediatric age
• Occupation or position held	PhD student
• Dates (from – to)	**2014-2019**
• Name and address of employer	Istituto Giannina Gaslini – Istituto Pediatrico di Ricovero e Cura a Carattere Scientifico, Via Gerolamo Gaslini 5, 16147, Genoa, Italy
• Type of business or sector	Pediatric residency
• Occupation or position held	Resident, Pediatrician
• Main activities and responsibilities	M.D.
• Dates (from – to)	**2011-2014**
• Name and address of employer	Azienda Ospedaliera Universitaria Federico II, Via Sergio Pansini 5, 80131, Naples, Italy
• Type of business or sector	Neurosurgery residency
• Occupation or position held	3rd year resident, resigned
• Main activities and responsibilities	M.D.

EDUCATION AND TRAINING

• Dates (from – to)	**2019-present**
• Name and type of organisation providing education and training	Università degli Studi di Genova
• Principal subjects/occupational skills covered	PhD: neuromuscular, neurometabolic, and neurodegenerative disorders in pediatric age
• Title of qualification awarded	PhD student

• Dates (from – to)	**2014-2019**
• Name and type of organisation providing education and training	Istituto Giannina Gaslini – Istituto Pediatrico di Ricovero e Cura a Carattere Scientifico, Via Gerolamo Gaslini 5, 16147, Genoa, Italy; Università Degli Studi di Genova, Via Balbi, 5, 16126, Genoa, Italy
• Principal subjects/occupational skills covered	Pediatric residency
• Title of qualification awarded	Pediatrician Dissertation: 'Whole Exome Sequencing in pediatric patients with intellectual disability: diagnostic impact and scientific implications.' Final vote: 50/50 cum laude
• Dates (from – to)	**July 2019-December 2019**
• Name and type of organisation providing education and training	Queen Square Institute of Neurology, University College of London (UCL) Queen Square, London WC1N 3BG
• Principal subjects/occupational skills covered	Next Generation Sequencing data analysis, gene discovery, functional studies
• Title of qualification awarded	Research Fellow (Prof. Henry Houlden Lab)
• Dates (from – to)	**February 2019-April 2019**
• Name and type of organisation providing education and training	Telethon Institute of Genetics and Medicine (TIGEM), Via Campi Flegrei, 34, 80078, Pozzuoli, Naples, Italy
• Principal subjects/occupational skills covered	Whole Exome Sequencing data interpretation in genetic disorders of unknown cause, in the context of Telethon Undiagnosed Disease Project (TUDP)
• Title of qualification awarded	Research collaborator
• Dates (from – to)	**May 2018-August 2018**
• Name and type of organisation providing education and training	The Montreal Children's Hospital McGill University Health Centre, 1001 Decarie Blvd, Montreal, QC H4A 3J1, Canada
• Principal subjects/occupational skills covered	Medical Genetics Observership
• Title of qualification awarded	Medical Genetics Observer
• Dates (from – to)	**2011-2014**
• Name and type of organisation providing education and training	Azienda Ospedaliera Universitaria Federico II, Via Sergio Pansini 5, 80131, Naples, Italy
• Principal subjects/occupational skills covered	Neurosurgery residency
• Title of qualification awarded	3rd year resident in Neurosurgery, *resigned*
• Dates (from – to)	**2011-2012**
• Name and type of organisation providing education and training	United States Medical Licensing Examination (USMLE), 3750 Market Street Philadelphia, PA 19104-3102
• Principal subjects/occupational skills covered	Step 1 USMLE: pass Step 2 CK USMLE: pass
• Dates (from – to)	**2005-2010**
• Name and type of organisation	Università degli Studi di Napoli Federico II,

providing education and training	Azienda Ospedaliera Universitaria Federico II, Via Sergio Pansini 5, 80131, Naples, Italy
• Principal subjects/occupational skills covered	Dissertation title: 'Endoscopic treatment of intraventricular tumors' Final vote: 110/110 cum laude.
• Title of qualification awarded	Honours Master Degree in Medicine and Surgery, M.D.

PERSONAL SKILLS AND COMPETENCES

MOTHER TONGUE	**ITALIAN**
OTHER LANGUAGES	
	ENGLISH
• Reading skills	VERY GOOD (C1)
• Writing skills	VERY GOOD (C1)
• Verbal skills	VERY GOOD (C1)
	FRENCH
• Reading skills	GOOD
• Writing skills	GOOD
• Verbal skills	GOOD
SOCIAL SKILLS AND COMPETENCES	STRONG DETERMINATION AND SELF-MOTIVATION ORGANIZATIONAL SKILLS IN TEAMWORK GOOD INTERPERSONAL COMMUNICATION SKILLS ABILITY TO ADAPT TO MULTICULTURAL ENVIRONMENTS
ORGANISATIONAL SKILLS AND COMPETENCES	SENSE OF ORGANISATION IN CLINICAL ACTIVITY AND RESEARCH RESEARCH COOPERATION WITH VARIOUS SCIENTIFIC PROFESSIONALS (CLINICIANS, SURGEONS, BIOLOGISTS, RADIOLOGISTS)
TECHNICAL SKILLS AND COMPETENCES	PROFICIENT WITH MICROSOFT OFFICE SUITE PROFICIENT WITH COMMON OPERATING SYSTEMS: WINDOWS, MAC, IOS, ANDROID
ARTISTIC SKILLS AND COMPETENCES	SINGING WRITING ELECTRIC GUITAR PLAYING
OTHER SKILLS AND COMPETENCES	PROVIDED GUIDANCE TO UNDERGRADUATE STUDENT IN THE PREPARATION OF GRADUATION THESIS ('ENDOSCOPY IN THE TREATMENT OF INTRACRANIAL CYSTS')
DRIVING LICENCE(S)	Category B

SPEAKER

25th-27th September 2014, Vicenza (Venice), Italy, 63th national congress of Italian Neurosurgery Society (SiNch): 'ETV in idiopathic normal pressure hydrocephalus: indications and clinical results.' (report on 'ETV nell'idrocefalo normoteso idiopatico: indicazioni e risultati clinici.')

16th-19th June 2018, Milan, Italy, European Society of Human Genetics (ESHG) Conference: 'A novel pathogenic MYH3 mutation in a child with Sheldon-Hall syndrome and vertebral fusions.'

6th-9th June 2020, 2020 European Society of Human Genetics (ESHG) Virtual Conference 2020: 'Biallelic MFSD2A variants associated with congenital microcephaly, developmental delay, and recognizable neuroimaging features.'

27th-30th October 2020, American Society of Human Genetics (ASHG), virtual meeting 2020: 'Biallelic variants in ADARB1, encoding a dsRNA-specific adenosine deaminase, cause a severe developmental and epileptic encephalopathy.'

9th-11th June 2021, 44° Italian congress of the Italian League Against Epilepsy (LICE) 2021: 'Genotype-phenotype spectrum of the developmental and epileptic encephalopathy caused by inosine triphosphatase deficiency.' ('Spettro genotipico e fenotipico della encefalopatia epilettica e dello sviluppo causata da deficit di inosina trifosfato pirofosfatasi')

18th-22nd October 2021, American Society of Human Genetics (ASHG), virtual meeting 2021: 'Biallelic loss-of-function variants in CACHD1, encoding an α2δ-like voltage-gated calcium channel regulator, cause a neurodevelopmental, craniofacial, and genitourinary syndrome.'

25th-27th November 2021, Italian Society of Pediatric Neurology (SINP), national conference 2021: 'RAC3 variants impair axon guidance and disrupt intracortical neuronal migration, leading to heterogeneous neurodevelopmental phenotypes.'

11th-14th June 2022, European Society of Human Genetics (ESHG) Conference 2022: 'Variant-specific changes in RAC3 function disrupt corticogenesis in neurodevelopmental phenotypes.'

PUBLICATIONS (N=69)
(CHRONOLOGICAL ORDER)
(EXCLUDING ERRATA)

Atypical choroid plexus papilloma: spontaneous resolution of diffuse leptomeningeal contrast enhancement after primary tumor removal in 2 pediatric cases. Scala M, Morana G, Milanaccio C, Pavanello M, Nozza P, Garrè ML. *J Neurosurg Pediatr***.** 2017 Sep;20(3):284-288. doi: 10.3171/2017.2.PEDS16526. Epub 2017 Jul 7.

A novel pathogenic MYH3 mutation in a child with Sheldon-Hall syndrome and vertebral fusions. Marcello Scala, Andrea Accogli, Elisa De Grandis, Anna Allegri, Christoph P. Bagowski, Moneef Shoukier, Mohamad Maghnie, Valeria Capra. *Am J Med Genet A*. 2018 Jan 5. doi: 10.1002/ajmg.a.38593. [Epub ahead of print]

When and why is surgical revascularization indicated for the treatment of moyamoya syndrome in patients with RASopathies? A systematic review of the literature and a single institute experience. Scala M, Fiaschi P, Capra V, Garrè ML, Tortora D, Ravegnani M, Pavanello M. *Childs Nerv Syst*. 2018 Jul;34(7):1311-1323. doi: 10.1007/s00381-018-3833-7. Epub 2018 May 24.

CNNM2 homozygous mutations cause severe refractory hypomagnesemia, epileptic encephalopathy and brain malformations. Accogli A, Scala M, Calcagno A, Napoli F, Di Iorgi N, Arrigo S, Mancardi MM, Prato G, Pisciotta L, Nagel M, Severino M, Capra V. *Eur J Med Genet*. 2019 Mar;62(3):198-203. doi: 10.1016/j.ejmg.2018.07.014. Epub 2018 Jul 17.

Novel CNS malformations and skeletal anomalies in a patient with Beaulieu-Boycott-Innes syndrome. Accogli A, Scala M, Calcagno A, Castello R, Torella A, Musacchia F, Allegri AME, Mancardi MM, Maghnie M, Severino M; Telethon Undiagnosed Diseases Program, Nigro V, Capra V. *Am J Med Genet A*. 2018 Dec;176(12):2835-2840. doi: 10.1002/ajmg.a.40534. Epub 2018 Sep 20.

Loss of tubulin deglutamylase CCP1 causes infantile-onset neurodegeneration. Shashi V, Magiera MM, Klein D, Zaki M, Schoch K, Rudnik-Schöneborn S, Norman A, Lopes Abath Neto O, Dusl M, Yuan X, Bartesaghi L, De Marco P, Alfares AA, Marom R, Arold ST, Guzmán-Vega FJ, Pena LD, Smith EC, Steinlin M, Babiker MO, Mohassel P, Foley AR, Donkervoort S, Kaur R, Ghosh PS, Stanley V, Musaev D, Nava C, Mignot C, Keren B, Scala M, Tassano E, Picco P, Doneda P, Fiorillo C, Issa MY, Alassiri A, Alahmad A, Gerard A, Liu P, Yang Y, Ertl-Wagner B, Kranz PG, Wentzensen IM, Stucka R, Stong N, Allen AS, Goldstein DB; Undiagnosed Diseases Network, Schoser B, Rösler KM, Alfadhel M, Capra V, Chrast R, Strom TM, Kamsteeg EJ, Bönnemann CG, Gleeson JG, Martini R, Janke C, Senderek J. *EMBO J*. 2018 Dec 3;37(23). pii: e100540. doi: 10.15252/embj.2018100540. Epub 2018 Nov 12.

Familial ROBO1 deletion associated with ectopic posterior pituitary, duplication of the pituitary stalk and anterior pituitary hypoplasia. Scala M, Accogli A, Allegri AME, Tassano E, Severino M, Morana G, Maghnie M, Capra V. *J Pediatr Endocrinol Metab*. 2018 Dec 11. pii: /j/jpem.ahead-of-print/jpem-2018-0272/jpem-2018-0272.xml. doi: 10.1515/jpem-2018-0272.

Radiation-induced moyamoya syndrome after proton therapy in a child with clival craniopharyngioma: natural history and surgical treatment. Scala M, Vennarini S, Garrè ML, Tortora D, Cianchetti M, Fellin F, Lorentini S, Pavanello M. *World Neurosurg*. 2018 Dec 21. pii: S1878-8750(18)32871-7. doi: 10.1016/j.wneu.2018.12.048.

Three de novo DDX3X variants associated with distinctive brain developmental abnormalities and brain tumor in intellectually disabled females. Scala M*, Torella A*, Severino M, Morana G, Castello R, Accogli A, Verrico A, Vari MS, Cappuccio G, Pinelli M, Vitiello G, Terrone G, D'Amico A; TUDP consortium, Nigro V, Capra V. *Eur J Hum Genet*. 2019 Apr 1. doi: 10.1038/s41431-019-0392-7.

Pelizaeus-Merzbacher Disease due to PLP1 frameshift mutation in a female with nonrandom skewed X-chromosome inactivation. Marcello Scala, Monica Traverso, Valeria Capra, Maria Stella Vari, Mariasavina Severino, Serena Grossi, Federico Zara, Pasquale Striano, Carlo Minetti. *Neuropediatrics*. 2019 May 28. doi: 10.1055/s-0039-1688954.

Abnormal circadian rhythm in patients with GRIN1-related developmental epileptic encephalopathy. Scala M, Amadori E, Fusco L, Marchese F, Capra V, Minetti C, Vari MS, Striano P. *European Journal of Paediatric Neurology*. doi: 10.1016/j.ejpn.2019.05.011.

Aggressive desmoid fibromatosis in Kabuki syndrome: expanding the tumor spectrum. Marcello Scala, Giovanni Morana, Angela Rita Sementa, Giuseppe Merla, Gianluca Piatelli, Valeria Capra, Marco Pavanello. *Pediatr Blood Cancer*. 2019 May 27:e27831. doi: 10.1002/pbc.27831.

16p12.2 Microdeletion in a patient with autosomal recessive deafness-22 (DFNB22). Elisa Tassano, Patrizia Ronchetto, Annalisa Calcagno, Patrizia Fiorio, Giorgio Gimelli, Valeria Capra, Marcello Scala. *J Genet* (2019) 98: 56. https://doi.org/10.1007/s12041-019-1107-0

Novel homozygous TSFM pathogenic variant associated with encephalocardiomyopathy with sensorineural hearing loss and peculiar neuroradiologic findings. Marcello Scala*, Giorgia Brigati*, Chiara Fiorillo, Claudia Nesti, Anna Rubegni, Marina Pedemonte, Claudio Bruno, Mariasavina Severino, Maria Derchi, Carlo Minetti, F.M. Santorelli. *Neurogenetics* 2019. https://doi.org/10.1007/s10048-019-00582-5

AMPA Receptor GluA2 Subunit Defects Are a Cause of Neurodevelopmental Disorders. Salpietro V, Dixon CL, Guo H, SYNAPS study group, et al. *Nat Commun*. 2019;10(1):3094. Published 2019 Jul 12. doi:10.1038/s41467-019-10910-w

Biallelic Variants in CTU2 Cause DREAM-PL Syndrome and Impair Thiolation of tRNA Wobble U34. Shaheen R, Mark P, Prevost CT, Al Kindi A, Alhag A, Estwani F, Al-Sheddi T, Alobeid E, Alenazi MM, Ewida N, Ibrahim N, Hashem M, Abdulwahab F, Bryant EM, Spinelli E, Millichap J, Barnett SS, Kearney HM, Accogli A, Scala M, Capra V, Nigro V, Fu D, Alkuraya FS. *Hum Mutat*. 2019 Jul 13. doi: 10.1002/humu.23870. [Epub ahead of print]

Radiation-induced moyamoya syndrome in children with brain tumors: case series and literature review. Scala M, Fiaschi P, Cama A, Consales A, Piatelli G, Giannelli F, Barra S, Satragno C, Pacetti M, Secci F, Tortora D, Garrè ML, Pavanello M. *World Neurosurg*. 2019 Dec 2. pii: S1878-8750(19)33005-0. doi: 10.1016/j.wneu.2019.11.155. [Epub ahead of print]

Advances in genetic testing and optimization of clinical management in children and adults with epilepsy. Scala M, Bianchi A, Bisulli F, Coppola A, Elia M, Trivisano M, Pruna D, Pippucci T, Canafoglia L, Lattanzi S, Franceschetti S, Nobile C, Gambardella A, Michelucci R15, Zara F, Striano P. *Expert Rev Neurother*. 2020 Jan 27:119. doi:10.1080/14737175.2020. 1713101. [Epub ahead of print]

RSRC1 loss-of-function variants cause mild-to-moderate autosomal recessive intellectual disability. Marcello Scala, Majid Mojarrad, Saima Riazuddin, Karlla W. Brigatti, Zineb Ammous, Julie S. Cohen, Heba Hosny, Muhammad A. Usmani, Mohsin Shahzad, Sheikh Riazuddin, Valentina Stanley, Atiye Eslahi, Richard E. Person, Hasnaa M. Elbendary, Anne M. Comi, Laura Poskitt, Vincenzo Salpietro, Queen Square Genomics, Jill A. Rosenfeld, Katie B. Williams, Dana Marafi, Fan Xia, Marta Biderman Waberski, Maha S. Zaki, Joseph Gleeson, Erik Puffenberger, Henry Houlden, Reza Maroofian. *Brain*. 2020 Apr 1;143(4):e31. doi: 10.1093/brain/awaa070.

Congenital posterior cervical spine malformation due to biallelic c.240-4T>G RIPPLY2 variant: a discrete entity. Margaux Serey-Gaut*, Marcello Scala*, Bruno Reversade, Lyse Ruaud, Christelle Cabrol, Francesco Musacchia, Annalaura Torella, Andrea Accogli, Nathalie Escande-Beillard, Jean Langlais, Gianluca Piatelli, Alessandro Consales, Vincenzo Nigro, Valeria Capra, Lionel Van Maldergem. *Am J Med Genet A*. 2020, Jun;182(6):1466-1472. doi: 10.1002/ajmg.a.61549. Epub 2020 Mar 25.

Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. Caroline Neuray, Reza Maroofian, Marcello Scala, Tipu Sultan, Gurpur S. Pai, Majid Mojarrad, Heba El Khashab, Leigh deHoll, Wyatt Yue, Hessa S. Alsaif, Maria N. Zanetti, Oscar Bello, Richard Person, Atieh Eslahi, Zaynab Khazaei, Masoumeh H. Feizabadi, Stephanie Efthymiou, Hala T. El-Bassyouni, Doaa R. Soliman, Vincenzo Salpietro, Yalda Jamshidi, Fowzan S. Alkuraya, Henry Houlden*, Queen Square Genomics, SYNaPS Study Group. *Brain*, 2020 Aug 1;143(8):2388-2397. doi: 10.1093/brain/awaa178. PMID: 32705143; PMCID: PMC7447512.

Novel likely disease-causing CLN5 variants identified in Pakistani patients with neuronal ceroid lipofuscinosis. Azad B, Efthymiou S, Sultan T, Marcello Scala, Javeria Raza Alvi, Caroline Neuray, Natalia Dominik, SYNaPS Study Group; Asma Gul, Henry Houlden. *J Neurol Sci*. 2020;414:116826. doi:10.1016/j.jns.2020.116826.

Loss of Wwox perturbs neuronal migration and impairs early cortical development. Michele Iacomino*, Simona Baldassari*, Yuki Tochigi*, Katarzyna Kośla, Francesca Buffelli, Annalaura Torella, Mariasavina Severino, Dario Paladini, Luana Mandarà, Antonella Riva, Marcello Scala, Ganna Balagura, Andrea Accogli, Vincenzo Nigro, Carlo Minetti, Ezio Fulcheri, Federico Zara, Andrzej K. Bednarek, Pasquale Striano, Hiroetsu Suzuki, Vincenzo Salpietro. *Frontiers in Molecular Neuroscience*, 2020 Jun 11;14:644. doi: 10.3389/fnins.2020.00644. eCollection 2020.

Sinus pericranii, skull defects, and structural brainanomalies in TRAF7-related disorder. Andrea Accogli, Marcello Scala, Marco Pavanello, Mariasavina Severino, Carlo Gandolfo, Patrizia De Marco, Francesco Musacchia, Annalaura Torella, Michele Pinelli, Vincenzo Nigro, Valeria Capra. Birth Defects Res. 2020 May 27. doi: 10.1002/bdr2.1711.

Biallelic MFSD2A variants associated with congenital microcephaly, developmental delay, and recognizable neuroimaging features. Marcello Scala*, Geok Lin Chua*, Cheen Fei Chin, Hessa S Alsaif, Borovikov Artem, Saima Riazuddin, Sheikh Riazuddin, M. Chiara Manzini, Mariasavina Severino, Alvin Kuk, Hao Fan, Yalda Jamshidi, Mehran Beiraghi Toosi, Mohammad Doosti, Ehsan Ghayoor Karimiani, Vincenzo Salpietro, Elena Dadali, Galina Baydakova, Fedor Konovalov, Ekaterina Lozier, Emer O'Connor, Yasser Sabr, Abdullah Alfaifi, Farah Ashrafzadeh, Pasquale Striano, Federico Zara, Fowzan S Alkuraya, Henry Houlden, Reza Maroofian, David L. Silver. *Eur J Hum Genet*, In production.

Biallelic variants in ADARB1, encoding a dsRNA-specific adenosine deaminase, cause a severe developmental and epileptic encephalopathy. Reza Maroofian, Jiří Sedmík, Neda Mazaheri, Marcello Scala, Maha Zaki, Liam P. Keegan, Reza Azizi Malmiri, Mahmoud Issa, Gholamreza Shariati, Alireza Sedaghat, Christian Beetz, Peter Bauer, Hamid Galehdari, Mary A. O'Connell, Henry Houlden. *J Med Genet*, 2020 Jul 27;jmedgenet-2020-107048. doi: 10.1136/jmedgenet-2020-107048. Online ahead of print.

Structural brain anomalies in Cri-du-Chat syndrome: MRI findings in 14 patients and

possible genotype-phenotype correlations. Villa R, Fergnani VGC, Silipigni R, Guerneri S, Cinnante C, Guala A, Danesino C, Scola E, Conte G, Fumagalli M, Gangi S, Colombo L, Picciolini O, Ajmone PF, Accogli A, Madia F, Tassano E, Scala M, Capra V, Srour M, Spaccini L, Righini A, Greco D, Castiglia L, Romano C, Bedeschi MF. *Eur J Paediatr Neurol*. 2020 Jul 28:S1090-3798(20)30143-4. doi: 10.1016/j.ejpn.2020.07.002. Epub ahead of print. PMID: 32800423.

Spatial coefficient of variation applied to arterial spin labeling MRI may contribute to predict surgical revascularization outcomes in pediatric moyamoya vasculopathy. Tortora D, Scavetta C, Rebella G, Marta Bertamino, Marcello Scala, Thea Giacomini, Giovanni Morana, Marco Pavanello, Andrea Rossi, Mariasavina Severino. *Neuroradiology*. 2020;10.1007/s00234-020-02446-4. doi:10.1007/s00234-020-02446-4.

A relatively common homozygous TRAPPC4 splicing variant is associated with an early-infantile neurodegenerative syndrome. Ghosh SG*, Scala M*, Beetz C, Helman G, Stanley V, Yang X, Breuss MW, Mazaheri N, Selim L, Hadipour F, Pais L, Stutterd CA, Karageorgou V, Begtrup A, Crunk A, Juusola J, Willaert R, Flore LA, Kennelly K, Spencer C, Brown M, Trapane P, Hurst ACE, Lane Rutledge S, Goodloe DH, McDonald MT, Shashi V, Schoch K; Undiagnosed Diseases Network, Tomoum H, Zaitoun R, Hadipour Z, Galehdari H, Pagnamenta AT, Mojarrad M, Sedaghat A, Dias P, Quintas S, Eslahi A, Shariati G, Bauer P, Simons C, Houlden H, Issa MY, Zaki MS, Maroofian R, Gleeson JG. *Eur J Hum Genet*. 2020 Sep 8. doi: 10.1038/s41431-020-00717-5. Epub ahead of print. PMID: 32901138.

De novo ARHGEF9 missense variants associated with neurodevelopmental disorder in females: expanding the genotypic and phenotypic spectrum of ARHGEF9 disease in females. Scala M, Zonneveld-Huijssoon E, Brienza M, Mecarelli O, van der Hout AH, Zambrelli E, Turner K, Zara F, Peron A, Vignoli A, Striano P. *Neurogenetics*. 2020 Sep 17. doi: 10.1007/s10048-020-00622-5. Epub ahead of print. PMID: 32939676.

Limits and pitfalls of indirect revascularization in moyamoya disease and syndrome. Fiaschi P, Scala M, Piatelli G, Tortora D, Secci F, Cama A, Pavanello M. *Neurosurg Rev*. 2020 Sep 21. doi: 10.1007/s10143-020-01393-1. Epub ahead of print. PMID: 32959193.

Targeted re-sequencing for early diagnosis of genetic causes of childhood epilepsy: the Italian experience from the 'beyond epilepsy' project. Amadori E, Scala M, Cereda GS, Vari MS, Marchese F, Di Pisa V, Mancardi MM, Giacomini T, Siri L, Vercellino F, Serino D, Orsini A, Bonuccelli A, Bagnasco I, Papa A, Minetti C, Cordelli DM, Striano P. *Ital J Pediatr*. 2020 Jul 6;46(1):92. doi: 10.1186/s13052-020-00860-1. PMID: 32631363; PMCID: PMC7339579.

Letter to the Editor Regarding "Primary Aneurysmal Bone Cyst of the Thoracic Spine: A Pediatric Case Report". Balestrino A, Scala M, Fiaschi P, Piatelli G, Pavanello M. *World Neurosurg*. 2020 Dec;144:322. doi: 10.1016/j.wneu.2020.08.065. PMID: 33227864.

Expanding the phenotype of PIGS-associated early onset epileptic developmental encephalopathy. Efthymiou S, Dutra-Clarke M, Maroofian R, Kaiyrzhanov R, Scala M, Reza Alvi J, Sultan T, Christoforou M, Tuyet Mai Nguyen T, Mankad K, Vona B, Rad A, Striano P, Salpietro V, Guillen Sacoto MJ, Zaki MS, Gleeson JG, Campeau PM, Russell BE, Houlden H. *Epilepsia*. 2021 Feb;62(2):e35-e41. doi: 10.1111/epi.16801. Epub 2021 Jan 7. PMID: 33410539; PMCID: PMC7898547.

Italian cohort of Lafora disease: Clinical features, disease evolution, and genotype-phenotype correlations. Antonella Riva, Alessandro Orsini, Marcello Scala, … , Pasquale Striano, on behalf of Italian League Against Epilepsy Genetic Commission. *Journal of Neurological Sciences*. Published March 20, 2021 DOI:https://doi.org/10.1016/j.jns.2021.117409.

Electroclinical features of MEF2C haploinsufficiency-related epilepsy: A multicenter European study. Raviglione F, Douzgou S, Scala M, Mingarelli A, … , Striano P. *Seizure*. 2021 Mar 30;88:60-72. doi: 10.1016/j.seizure.2021.03.025. Epub ahead of print. PMID: 33831796.

Temporal-parietal-occipital epilepsy in GEFS+ associated with SCN1A mutation. Riva A, Coppola A, Balagura G, Scala M, Iacomino M, Marchese F, Amadori E, Lattanzi S, Meo R, Striano S, Salpietro V, Zara F, Minetti C, Striano P, Bilo L. *Epileptic Disord*. 2021 Apr 12. doi: 10.1684/epd.2021.1266. Epub ahead of print. PMID: 33851920.

Homozygous SCN1B variants causing early infantile epileptic encephalopathy 52 affect voltage-gated sodium channel function. Scala M, Efthymiou S, Sultan T, De Waele J, Panciroli M, Salpietro V, Maroofian R, Striano P, Van Petegem F, Houlden H, Bosmans F. *Epilepsia*. 2021 Apr 26. doi: 10.1111/epi.16913. Epub ahead of print. PMID: 33901312.

Genotype-Phenotype Correlations in Neurofibromatosis Type 1: A Single-Center Cohort Study. Scala M, Schiavetti I, Madia F, Chelleri C, Piccolo G, Accogli A, Riva A, Salpietro V, Bocciardi R, Morcaldi G, Di Duca M, Caroli F, Verrico A, Milanaccio C, Viglizzo G, Traverso M, Baldassari S, Scudieri P, Iacomino M, Piatelli G, Minetti C, Striano P, Garrè ML, De Marco P, Diana MC, Capra V, Pavanello M, Zara F. *Cancers (Basel)*. 2021 Apr 14;13(8):1879. doi: 10.3390/cancers13081879. PMID: 33919865; PMCID: PMC8070780.

Biallelic Variants in KIF17 Associated with Microphthalmia and Coloboma Spectrum. Riva A, Gambadauro A, Dipasquale V, Casto C, Ceravolo MD, Accogli A, Scala M, Ceravolo G, Iacomino M, Zara F, Striano P, Cuppari C, Di Rosa G, Cutrupi MC, Salpietro V, Chimenz R. *Int J Mol Sci*. 2021 Apr 25;22(9):4471. doi: 10.3390/ijms22094471. PMID: 33922911.

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Genotype-phenotype spectrum and correlations in Xia-Gibbs syndrome: report of five novel cases and literature review. Ferruccio Romano, Mariateresa Falco, Gerarda Cappuccio, …, Emanuele Agolini, Antonio Novelli, Livia Garavelli, TUDP, Pasquale Striano, Gioacchino Scarano, Vincenzo Nigro, Marcello Scala*, Valeria Capra*. *Birth Defects Res*. 2022 Jun 18. doi: 10.1002/bdr2.2058. Epub ahead of print. PMID: 35716097.

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ADGRL1 haploinsufficiency causes a variable spectrum of neurodevelopmental disorders in humans and in a mouse model with altered synaptic activity. Antonio Vitobello, Benoit Mazel, Vera G Lelianova, Jason Suckling, …, Marcello Scala, Quentin Thomas, Serguei V Kozlov, Laurence Faivre, Christel Thauvin-Robinet, and Yuri Ushkaryov. Accepted for publication in *Am J Hum Genet*. 2022. AJHG-D-22-00227

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(UPDATED TO 09/22)

Scopus: **11** (https://www.scopus.com/authid/detail.uri?authorId=57195634402)
Google Scholar: **12** (https://scholar.google.it/citations?user=ofLKoQ4AAAAJ&hl=it)
i10-index (Google Scholar): **18** (https://scholar.google.it/citations?user=ofLKoQ4AAAAJ&hl=it)
Web of Science (ResearcherID ABE-7934-2020): **11**
Researchgate: **12** (https://www.researchgate.net/profile/Marcello-Scala/scores)

CITATIONS
(UPDATED TO 09/22)

Scopus: **600** (https://www.scopus.com/authid/detail.uri?authorId=57195634402)
Google Scholar: **919** (https://scholar.google.it/citations?user=ofLKoQ4AAAAJ&hl=it)
Web of Science (ResearcherID ABE-7934-2020): **551**
Researchgate: **801** (https://www.researchgate.net/profile/Marcello-Scala/scores)

STUDY GROUPS

Epi25 Collaborative in
Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. Epi25 Collaborative. Electronic address: jm4279@cumc.columbia.edu; Epi25 Collaborative. *Am J Hum Genet*. 2021 Apr 28:S0002-9297(21)00140-3.

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Mapping the human genetic architecture of COVID-19. COVID-19 Host Genetics Initiative. *Nature*. 2021 Jul 8. doi: 10.1038/s41586-021-03767-x. Epub ahead of print. PMID: 34237774.

REVIEWER ACTIVITY

Reviewer for the following journals:
- *Journal of Medical Genetics*
- *Clinical Genetics*
- *Molecular Neurobiology*
- *Neurological Sciences*
- *Molecular Genetics & Genomic Medicine*
- *Epilepsy Research*
- *Epilepsia Open*
- *Epileptic Disorders*

EDITORIAL TASKS

- Guest Editor for *Journal of Translational Genetics and Genomics*, Special issue 'Genetic Neurodevelopmental Diseases'
- Review Editor for *Frontiers in Neurology and Frontiers in Pediatrics*

TEACHING ACTIVITY

Seminars in pediatric neurology at the Gaslini Children's Hospital – University of Genoa (2020-2022)

AWARDS

'Best research presentation award' at the 63th national congress of *Italian Neurosurgery Society (SiNch)*, 25th-27th September 2014, Vicenza (Venice), Italy

Erasmus+ Traineeship, 2019-2020, *University of Genoa*, at Queen Square Institute of Neurology, University College of London (UCL), London, UK (Tutor: Prof. Henry Houlden)

'Best pediatric specialization thesis on rare diseases', 2019, *Società Italiana Malattie Genetiche Pediatriche e Disabilità Congenite SIMGePeD*, 'Whole Exome Sequencing in pediatric patients with intellectual disability: diagnostic impact and scientific implications.'

Erasmus+ Traineeship, 2020-2021, *University of Genoa*, at Queen Square Institute of Neurology, University College of London (UCL), London, UK (Tutor: Prof. Henry Houlden)

'Best oral research communication', 2021, Italian Society of Pediatric Neurology: 'RAC3 variants impair axon guidance and disrupt intracortical neuronal migration, leading to heterogeneous neurodevelopmental phenotypes.'

FIELDS OF STUDY

NGS data analysis, genomics, gene discovery

Clinical applications of Next Generation Sequencing (NGS) in Mendelian diseases through Whole Exome Sequencing (WES) and Whole Genome Sequencing (WGS)

Study of the genetic aspects of neurological disorders in childhood

Study of congenital central and peripheral nervous system abnormalities, with focus on developmental brain anomalies and neural tube defects

Study of the genetic aspects of peripheral and central nervous system tumors and neurocutaneous syndromes

Interest in the study of the clinical applications of gene therapy

Genova 21/10/2022