EUROPEAN CURRICULUM VITAE FORMAT

PERSONAL INFORMATION

Name	**SCALA MARCELLO**
Address	**6, Via del Commercio, 16167, Genoa, Italy**
Telephone	**Mobile: +39 3756906008 Home: +39 3756906008**
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Nationality	Italian
Place and date of birth	NAPLES, 19 OCTOBER 1987

WORK EXPERIENCE

• Dates (from – to)	**2023-present**
• Name and address of employer	Università degli Studi di Genova (DINOGMI)
• Type of business or sector	Researcher in Medical Genetics and Genomics
• Occupation or position held	Researcher
• Dates (from – to)	**2019-2022**
• 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)	**2023-present**
• Name and address of employer	Università degli Studi di Genova
• Type of business or sector	Researcher in Medical Genetics and Genomics
• Occupation or position held	Researcher
	2019-2022
	Università degli Studi di Genova
	PhD: neuromuscular, neurometabolic, and neurodegenerative disorders in pediatric age: - Implementation of knowledge on genomic data analysis and gene discovery approaches;

• Dates (from – to)	- Acquisition of competencies in the coordination of studies investigating the pathophysiological mechanisms underlying neurodevelopmental disorders;
• Name and type of organisation providing education and training	- Organization and coordination of several international collaborative studies in the field of medical genetics and genomics;
• Principal subjects/occupational skills covered	- Implementation of networking with leader international genomic and translational research centers and institutions (Aichi Human Service Center, BCM, CHOP, INSERM, TMI, UCL);
• Title of qualification awarded	- Acquisition of knowledge in basic statistical approaches to study genotype/phenotype correlations and genetics of polygenic conditions PhD
• 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 Pediatrician
• Title of qualification awarded	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 (Prof. Henry Houlden Lab), University College of London (UCL), Queen Square, London WC1N 3BG
• Principal subjects/occupational skills covered	<u>Next Generation Sequencing data analysis, gene discovery, functional studies:</u> - Acquisition of in-depth competencies in genomic data analysis towards the identification of novel candidate genes causing neurodevelopmental disorders and rare syndromes; - Acquisition of competencies in the organization and coordination of collaborative translational research studies involving genomic, clinical, and functional investigation; - Acquisition of competencies to perform basic molecular experiments, including Polymerase Chain Reaction (PCR), Gel electrophoresis, and Western Blot
• 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	<u>Genomic data analysis</u> in genetic disorders of unknown cause, in the context of Telethon Undiagnosed Disease Project (TUDP), under the supervision of Prof. Vincenzo Nigro (Università degli Studi della Campania "Luigi Vanvitelli" di Napoli): - Knowledge of the bioinformatic processing of raw exome/genome sequencing data; - Competency in the use of *in silico* tools to analyze exonic and genomic variants; - Competency in the analysis of genotype-phenotype correlations
• Title of qualification awarded	Research collaborator* (<u>This collaboration is currently still ongoing within the TUDP</u>)
• 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 (McGill University), 1001 Decarie Blvd, Montreal, QC H4A 3J1, Canada
• Principal subjects/occupational skills covered	<u>Medical Genetics Observership in the Department of Medical Genetics (Prof. Laura Russel):</u> - Mastery of the medical genetic evaluation of pediatric and adult patients; - Acquisition of the competencies necessary to counsel patients and report genetic findings; - Improvement of language skills in English and French <u>Molecular Genetics Observership in Prof. Andrea Ruchon's Lab:</u> - Acquisition of competency in the interpretation of genetic testing results (DNA/RNA sequencing, methylation analysis) - Acquisition of knowledge in the methods and technical/interpretation pitfalls of genetic tests (DNA/RNA sequencing, methylation analysis)
• 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: - Competency in the clinical and surgical management of adult and pediatric patients with brain malformations, brain tumors, spinal tumors, and discopathy - Acquisition of basic practical surgical skills
• 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)	**2005-2011**
• Name and type of organisation providing education and training	Università degli Studi di Napoli Federico II, 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 (C2)
• Writing skills	VERY GOOD (C2)
• Verbal skills	VERY GOOD (C2)
	FRENCH
• Reading skills	GOOD (B2)
• Writing skills	GOOD (B2)
• Verbal skills	GOOD (B2)
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

PROFESSIONAL ACHIEVEMENTS

SPEAKER
(ORAL PRESENTATION = 16)
(E-POSTER = 6)
(INVITED = 2)

- **25th-27th September 2014, Vicenza (Venice), Italy, 63th national congress of Italian Neurosurgery Society (SiNch) (oral presentation)**: '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 (e-poster):** '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 (e-poster):** '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 (e-poster):** '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 (oral presentation):** '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 (e-poster):** '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 (oral presentation):** '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 (e-poster):** 'Variant-specific changes in RAC3 function disrupt corticogenesis in neurodevelopmental phenotypes.'
- **7th October 2022, Italian League Against Epilepsy (LICE), Riunione annual Liguria, Piemonte e Valle d'Aosta (oral presentation):** 'Epilettologia in età evolutive: medicina di precisione'.
- **20th-22nd October 2022, Italian Society of Pediatric Neurology (SINP), national conference 2022 (oral presentation) (*invited*):** 'Exome sequencing in developmental and epileptic encephalopathies: a bridge between diagnostics and research'.
- **25th-29th October 2022, American Society of Human Genetics (ASHG) Conference 2022 (oral presentation): '***De novo* and biallelic variants in R3HDM1, encoding an Encore-like RNA binding protein hosting the microRNA MiR-128-1, disrupt cortical development and lead to neurodevelopmental phenotypes and epilepsy'.
- **10th-13th June 2023, European Society of Human Genetics (ESHG) Conference 2023 (oral presentation):** 'De novo variants in DENND5B perturb intracellular vesicular trafficking and cause neurodevelopmental disorders with epilepsy and white matter abnormalities'.
- **13th-16th September 2023, EuroDysmorpho Conference 2023 (European Reference Network ERN-ITHACA) (oral presentation):** 'Biallelic loss-of-function variants in voltage-gated calcium channel regulator CACHD1 cause neuronal dysfunction and multisystem developmental abnormalities'.
- **11th-15th September 2023, University of Genova PhD course in Neuroscience, 2023 Summer School II edition (oral presentation):** 'Novel disease gene discovery approaches for Neuromuscular Disorders'.
- **4th-6th October 2023, XXVI Congresso Nazionale della Società Italiana di Genetica Umana (SIGU) 2023 (oral presentation):** '*De novo* variants in *DENND5B* perturb intracellular vesicular trafficking and cause neurodevelopmental disorders with epilepsy and white matter abnormalities'.
- **1st-5th November 2023, American Society of Human Genetics (ASHG) Conference 2023 (oral presentation): '***De novo* and selfish maternal variants in *SYTL4*, encoding a member of the synaptotagmin-like protein family, disrupt vesicle exocytosis and cause an incompletely penetrant X-linked neurobehavioral disorder with autism, epilepsy, and spasticity'.
- **30th November-2nd December 2023, Italian Society of Pediatric Neurology (SINP), national conference 2023 (oral presentation) (*invited*):** 'Neurodevelopmental disorders and epilepsy'.
- **30th November-2nd December 2023, Italian Society of Pediatric Neurology (SINP), national conference 2023 (oral presentation):** 'Biallelic loss-of-function variants in voltage-gated calcium channel regulator CACHD1 cause neuronal dysfunction and multisystem developmental abnormalities'.
- **1st-4th June 2024, European Society of Human Genetics (ESHG) Conference 2024 (oral presentation):** 'De novo missense variants in EIF3I cause a neurodevelopmental disorder with dysmorphism, short stature, and midline brain defects'.
- **2nd-4th October 2024, XXVII Congresso Nazionale della Società Italiana di Genetica Umana (SIGU) 2024 (oral presentation):** 'De novo missense variants in EIF3I cause a neurodevelopmental disorder with dysmorphism, short stature, and midline brain defects'.
- **5th-9th November 2024, American Society of Human Genetics (ASHG) Conference 2024 (poster presentation): '**Genetic variants in *DDX53* contribute to Autism Spectrum Disorder associated with the Xp22.11 locus.'.
- **19th December 2024, Course at Istituto CSS-Mendel:** 'La genetica delle epilessie e delle sindromi epilettiche: basi molecolari, test genetici ed implicazioni cliniche'
- **- ERN Ithaca (European Reference Network for Rare Malformation Syndromes, Intellectual and Other Neurodevelopmental Disorders),** webinar on 'Rho-GTPase in intellectual disability and neurodevelopmental disorders': 'RAC3-related disorders of cortical development in human neurodevelopmental phenotypes'

MEETING ORGANIZATION

- **4th August 2023, International conference at Giannina Gaslini Children's Hospital/University of Genoa (Italy):** 'Connecting the dots: genomics, neuroimaging, and pathophysiology in understanding neurodevelopmental disorders'. *International guest: Prof. Koh-ichi Nagata (Institute for Developmental Research, Aichi Human Service Center, Kasugai, Japan).*

- **17th May 2024, International conference at Giannina Gaslini Children's Hospital/University of Genoa (Italy):** 'Neurodevelopmental and neurodegenerative disorders: gene discovery and translational research'. *International guest: Prof. Kevin Vaughan (Department of Biological Sciences, University of Notre Dame, Indiana, USA).*

SELECTED PUBLICATIONS
AUTHORSHIP LEGEND:
NAME: FIRST; LAST
(* = SHARED)
✉ = CORRESPONDING

- **Variant-specific changes in RAC3 function disrupt corticogenesis in neurodevelopmental phenotypes.** Marcello Scala*, Masashi Nishikawa*, Hidenori Ito*, Hidenori Tabata, .., Valeria Capra, Gregory Costain and Koh-ichi Nagata. *Brain*. 2022 Jul 19:awac106. doi: 10.1093/brain/awac106. Epub ahead of print. PMID: 35851598.
- **Novel loss-of-function variants expand ABCC9-related intellectual disability and myopathy syndrome.** Efthymiou S*, Scala M*, Nagaraj V, …, Nichols CG, Smeland MF, McClenaghan C. *Brain*. 2024 Jan 13:awae010. doi: 10.1093/brain/awae010. Epub ahead of print. PMID: 38217872.
- **Biallelic ZBTB11 variants associated with complex neuropsychiatric phenotype featuring Tourette syndrome.** Scala M✉, De Grandis E, .., Zara F, Striano P. *Brain*. 2022 Sep 7:awac323. doi: 10.1093/brain/awac323. Epub ahead of print. PMID: 36068688.
- **RSRC1 loss-of-function variants cause mild-to-moderate autosomal recessive intellectual disability.** Marcello Scala, Majid Mojarrad, .., Henry Houlden, Reza Maroofian. *Brain*. 2020 Apr 1;143(4):e31. doi: 10.1093/brain/awaa070.
- **Genetic variants in DDX53 contribute to autism spectrum disorder associated with the Xp22.11 locus.** Scala M, Bradley CA, Howe JL, Trost B, Salazar NB, Shum C, Mendes M, …, Scherer SW. *Am J Hum Genet*. 2025 Jan 2;112(1):154-167. doi: 10.1016/j.ajhg.2024.11.003. Epub 2024 Dec 19. PMID: 39706195; PMCID: PMC11739878.
- **De novo variants in DENND5B cause a neurodevelopmental disorder.** Scala M, Tomati V, Ferla M, …, Pedemonte N, Zara F. *Am J Hum Genet*. 2024 Feb 16:S0002-9297(24)00033-8. doi: 10.1016/j.ajhg.2024.02.001. Epub ahead of print. PMID: 38387458.
- **Biallelic loss-of-function variants in CACHD1 cause a novel neurodevelopmental syndrome with facial dysmorphism and multisystem congenital abnormalities.** Scala M, Khan K, Beneteau C, Fox RG, …, Bézieau S, Davis EE, Wells MF. *Genet Med*. 2023 Dec 27;26(4):101057. doi: 10.1016/j.gim.2023.101057. Epub ahead of print. PMID: 38158856.
- **Homozygous SCN1B variants causing early infantile epileptic encephalopathy 52 affect voltage-gated sodium channel function.** Scala M, Efthymiou S, …, Houlden H, Bosmans F. *Epilepsia*. 2021 Jun;62(6):e82-e87. doi: 10.1111/epi.16913. Epub 2021 Apr 26. PMID: 33901312; PMCID: PMC8585727.
- **Biallelic MFSD2A variants associated with congenital microcephaly, developmental delay, and recognizable neuroimaging features.** Scala M, Chua GL, Chin CF, …, Silver DL. *Eur J Hum Genet*. 2020 Nov;28(11):1509-1519. doi: 10.1038/s41431-020-0669-x. Epub 2020 Jun 22. PMID: 32572202; PMCID: PMC7576150.
- **A relatively common homozygous TRAPPC4 splicing variant is associated with an early-infantile neurodegenerative syndrome.** Ghosh SG*, Scala M*, Beetz C, …, Gleeson JG. *Eur J Hum Genet*. 2021 Feb;29(2):271-279. doi: 10.1038/s41431-020-00717-5. Epub 2020 Sep 8. PMID: 32901138; PMCID: PMC7868361.
- **Three de novo DDX3X variants associated with distinctive brain developmental abnormalities and brain tumor in intellectually disabled females.** Scala M*✉, Torella A*, …, Capra V. *Eur J Hum Genet*. 2019 Aug;27(8):1254-1259. doi: 10.1038/s41431-019-0392-7. Epub 2019 Apr 1. PMID: 30936465; PMCID: PMC6777618.
- **De novo truncating NOVA2 variants affect alternative splicing and lead to heterogeneous neurodevelopmental phenotypes.** Scala M✉, Drouot N, MacLennan SC, .., Zara F, Striano P, Piton A. *Hum Mutat*. 2022 May 24. doi: 10.1002/humu.24414. Epub ahead of print. PMID: 35607920.
- **DAG1 haploinsufficiency is associated with sporadic and familial isolated or pauci-symptomatic hyperCKemia.** Traverso M, Baratto S, …, Fiorillo C, Scala M✉. *Eur J Hum Genet*. 2024 Jan 4. doi: 10.1038/s41431-023-01516-4. Epub ahead of print. PMID: 38177406.
- **Refining the electroclinical spectrum of NPRL3-related epilepsy: A novel multiplex family and literature review.** Dainelli A, Iacomino M, Rossato S, …, Zara F, Scala M*✉, Striano P*. *Epilepsia Open*. 2023 Jul 25. doi: 10.1002/epi4.12798. Epub ahead of print. PMID: 37491868.
- **Novel KIF26A variants associated with pediatric intestinal pseudo-obstruction (PIPO) and brain developmental defects.** Nosrati MSS, Doustmohammadi A, …, Müller A, Zara F, Capra V, Scala M✉. *Clin Genet*. 2024 Sep 21. doi: 10.1111/cge.14621. Epub ahead of print. PMID: 39305096.

ALL PUBLICATIONS
(N = 141, PUBMED)
(N = 139, SCOPUS)
(CHRONOLOGICAL ORDER)

AUTHORSHIP LEGEND:
NAME: FIRST (41); LAST 11)
(* = SHARED)
✉ = CORRESPONDING (28)

- **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, .., Scala M, .., 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, .., 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, .., 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, .., 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, .., 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, .., 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, .., 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, .., 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.
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- **Genetic variants in DDX53 contribute to Autism Spectrum Disorder associated with the Xp22.11 locus.** Scala M, Bradley CA, Howe JL, …, Scherer SW. *medRxiv* [Preprint]. 2023 Dec 27:2023.12.21.23300383. doi: 10.1101/2023.12.21.23300383. PMID: 38234782; PMCID: PMC10793518.
- **Variant-specific pathophysiological mechanisms of AFF3 differently influence transcriptome profiles.** Bassani S, Chrast J, Ambrosini G, …, Scala M, …, Guex N, Reymond A. *medRxiv* [Preprint]. 2024 Jan 17:2024.01.14.24301100. doi: 10.1101/2024.01.14.24301100. PMID: 38293053; PMCID: PMC10827271.
- **Surgical revascularization as a procedure to prevent neurological complications in children with moyamoya syndrome associated with neurofibromatosis I: a single institution case series.** Morello A, Scala M, .., Piatelli G, Pavanello M. *Childs Nerv Syst*. 2024 Feb 6. doi: 10.1007/s00381-024-06304-z. Epub ahead of print. PMID: 38316674.
- **Expanding the phenotype of UPF3B-related disorder: Case reports and literature review.** Romano F, Haanpää MK, …, Capra V, Scala M✉. *Am J Med Genet A*. 2024 Feb 6:e63534. doi: 10.1002/ajmg.a.63534. Epub ahead of print. PMID: 38318947.
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- **Expansion of the neurodevelopmental phenotype of individuals with EEF1A2 variants and genotype-phenotype study.** Paulet A, Bennett-Ness C, Ageorges F, …, Scala M, Abbott CM, Ruaud L. *Eur J Hum Genet*. 2024 Sep;32(9):1144-1149. doi: 10.1038/s41431-024-01560-8. Epub 2024 Feb 15.
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- **Variant-specific pathophysiological mechanisms of AFF3 differently influence transcriptome profiles.** Bassani S, Chrast J, …, Scala M, …, Reymond A. *Genome Med*. 2024 May 30;16(1):72. doi: 10.1186/s13073-024-01339-y. PMID: 38811945; PMCID: PMC11137988.
- **Structural variant calling and clinical interpretation in 6224 unsolved rare disease exomes.** Demidov G, Laurie S, …, Scala M, …, Ossowski S. *Eur J Hum Genet*. 2024 Aug;32(8):998-1004. doi: 10.1038/s41431-024-01637-4. Epub 2024 May 31. PMID: 38822122; PMCID: PMC11291474.
- **Novel causative variants in Legius syndrome: SPRED1 Genotype spectrum expansion.** Chelleri C, Brolatti N, …, Zara F, Scala M✉. *Am J Med Genet A*. 2024 Jul 19:e63824. doi: 10.1002/ajmg.a.63824. Epub ahead of print. PMID: 39031930.
- **Expanding the Mutational Landscape and Clinical Phenotype of CHD2-Related Encephalopathy.** Clara-Hwang A, Stefani S, Lau T, Scala M, …, Nicolaides P, Striano P, Efthymiou S. *Neurol Genet*. 2024 Jul 11;10(4):e200168. doi: 10.1212/NXG.0000000000200168. PMID: 39035822; PMCID: PMC11259532.
- **V-ATPase Dysfunction in the Brain: Genetic Insights and Therapeutic Opportunities.** Falace A, Volpedo G, Scala M, Zara F, Striano P, Fassio A. *Cells*. 2024 Aug 28;13(17):1441. doi: 10.3390/cells13171441. PMID: 39273013; PMCID: PMC11393946.
- **An interconnected data infrastructure to support large-scale rare disease research.** Johansson LF, Laurie S, …; Solve-RD consortium. *Gigascience*. 2024 Jan 2;13:giae058. doi: 10.1093/gigascience/giae058. PMID: 39302238; PMCID: PMC11413801.
- **Novel KIF26A variants associated with pediatric intestinal pseudo-obstruction (PIPO) and brain developmental defects.** Nosrati MSS, Doustmohammadi A, …, Müller A, Zara F, Capra V, Scala M✉. *Clin Genet*. 2024 Sep 21. doi: 10.1111/cge.14621. Epub ahead of print. PMID: 39305096.
- **Exome sequencing of 20,979 individuals with epilepsy reveals shared and distinct ultra-rare genetic risk across disorder subtypes.** Epi25 Collaborative. *Nat Neurosci*. 2024 Oct;27(10):1864-1879. doi: 10.1038/s41593-024-01747-8. Epub 2024 Oct 3. PMID: 39363051.
- **BCL11A intellectual developmental disorder: defining the clinical spectrum and genotype-phenotype correlations.** Peron A, D'Arco F, …, Scala M, …, Dias C. *Eur J Hum Genet*. 2024 Oct 24. doi: 10.1038/s41431-024-01701-z. Epub ahead of print. PMID: 39448799.
- **Comprehensive reanalysis for CNVs in ES data from unsolved rare disease cases results in new**

diagnoses. Demidov G, Yaldiz B,…; Solve-RD Consortium; Laurie S. *NPJ Genom Med*. 2024 Oct 26;9(1):49. doi: 10.1038/s41525-024-00436-6. PMID: 39461972; PMCID: PMC11513043.

- **HMGCS1 variants cause rigid spine syndrome amenable to mevalonic acid treatment in an animal model.** Dofash LNH, Miles LB, Saito Y, …, Scala M, …, Ravenscroft G. *Brain*. 2024 Nov 12:awae371. doi: 10.1093/brain/awae371. Epub ahead of print. PMID: 39531736.
- **Neurological and psychiatric phenotype of a multicenter cohort of patients with SETD5-related neurodevelopmental disorder.** De Falco A, De Dominicis A, …, Scala M, …, Terrone G. *Eur J Paediatr Neurol*. 2024 Nov 23;54:8-17. doi: 10.1016/j.ejpn.2024.11.008. Epub ahead of print. PMID: 39603091.
- **National survey on the prevalence of single-gene aetiologies for genetic developmental and epileptic encephalopathies in Italy.** Mei D, Balestrini S, …, Scala M, …, Guerrini R. *J Med Genet*. 2024 Dec 31;62(1):25-31. doi: 10.1136/jmg-2024-110328. PMID: 39613335; PMCID: PMC11877070.
- **The p.R66W Variant in RAC3 Causes Severe Fetopathy Through Variant-Specific Mechanisms.** Sugawara R, Ito H, Tabata H, Ueda H, Scala M, Nagata KI. *Cells*. 2024 Dec 9;13(23):2032. doi: 10.3390/cells13232032. PMID: 39682779; PMCID: PMC11640247.
- **Genetic variants in DDX53 contribute to autism spectrum disorder associated with the Xp22.11 locus.** Scala M, Bradley CA, Howe JL, …, Scherer SW. *Am J Hum Genet*. 2025 Jan 2;112(1):154-167. doi: 10.1016/j.ajhg.2024.11.003. Epub 2024 Dec 19. PMID: 39706195; PMCID: PMC11739878.
- **DNA-binding affinity and specificity determine the phenotypic diversity in BCL11B-related disorders.** Lessel I, Baresic A, Chinn IK, …, Scala M, …, Lessel D. *Am J Hum Genet*. 2025 Feb 6;112(2):394-413. doi: 10.1016/j.ajhg.2024.12.012. Epub 2025 Jan 10. PMID: 39798569; PMCID: PMC11866971.
- **Variants in Chromatin Remodeling Genes Are Involved in Patients With Chiari Malformation Type 1.** Romano F, Cerminara M, …, Scala M, …, Capra V. *Birth Defects Res*. 2025 Feb;117(2):e2446. doi: 10.1002/bdr2.2446. PMID: 39907171; PMCID: PMC11795737.
- **Novel De Novo RALA Missense Variants Expand the Genotype Spectrum of Hiatt-Neu-Cooper Neurodevelopmental Syndrome.** Dainelli A, Nosrati MSS, …, Zara F, Scala M. *Mol Genet Genomic Med*. 2025 Feb;13(2):e70072. doi: 10.1002/mgg3.70072. PMID: 39918382; PMCID: PMC11803908.
- **CDK13-Related Disorder: Novel Insights From A Series of 27 Cases and Recommendations for Clinical Management.** Contrò G, Baroni MC, Caraffi SG, …, Scala M, …, Garavelli L. *Clin Genet*. 2025 Feb 19. doi: 10.1111/cge.14726. Epub ahead of print. PMID: 39971730.
- **A p.N92K variant of the GTPase RAC3 disrupts cortical neuron migration and axon elongation.** Sugawara R, Hamada K, Ito H, Scala M, …, Nagata KI. *J Biol Chem*. 2025 Feb 25:108346. doi: 10.1016/j.jbc.2025.108346. Epub ahead of print. PMID: 40015633.
- **Zinc transporter ZIP13 G289R variant from Spondylocheirodysplastic Ehlers-Danlos syndrome (SCD-EDS) is associated with abnormal hair quality.** Brito S, Park G, Lee GH, …, Scala M, Bin BH. *J Invest Dermatol*. 2025 Feb 26:S0022-202X(25)00294-5. doi: 10.1016/j.jid.2025.02.133. Epub ahead of print. PMID: 40020993.

BOOK CHAPTERS

- **CT Scan Data Analysis in Malformations of Cortical Development.** Scala M, Severino M. *Methods Mol Biol*. 2024;2794:271-280. doi: 10.1007/978-1-0716-3810-1_22. PMID: 38630236.
- **MRI Data Analysis in Malformations of Cortical Development.** Severino M, Tortora D, Scala M. *Methods Mol Biol*. 2024;2794:281-292. doi: 10.1007/978-1-0716-3810-1_23. PMID: 38630237.

H-INDEX (UPDATED TO 03/25)

Scopus: **23** (https://www.scopus.com/authid/detail.uri?authorId=57195634402)
Google Scholar: **25** (https://scholar.google.it/citations?user=ofLKoQ4AAAAJ&hl=it)
i10-index (Google Scholar): **46** (https://scholar.google.it/citations?user=ofLKoQ4AAAAJ&hl=it)
Web of Science (ResearcherID ABE-7934-2020): **23**

CITATIONS (UPDATED TO 03/25)

Scopus: **2,094** (https://www.scopus.com/authid/detail.uri?authorId=57195634402)
Google Scholar: **2732** (https://scholar.google.it/citations?user=ofLKoQ4AAAAJ&hl=it)
Web of Science (ResearcherID ABE-7934-2020): **2,357**

STUDY GROUPS

- Epi25 Collaborative in:

- Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. Epi25 Collaborative. *Am J Hum Genet*. 2021 Apr 28:S0002-9297(21)00140-3.
- Genome-wide identification and phenotypic characterization of seizure-associated copy number variations in 741,075 individuals. *Nat Commun*. 2023 Jul 20;14(1):4392.
- Exome sequencing of 20,979 individuals with epilepsy reveals shared and distinct ultra-rare genetic risk across disorder subtypes. Epi25 Collaborative. *Nat Neurosci*. 2024 Oct;27(10):1864-1879.

- COVID-19 Host Genetics Initiative in:

- Mapping the human genetic architecture of COVID-19. COVID-19 Host Genetics Initiative. *Nature*. 2021 Jul 8.

- Solve-RD Consortia in:

- A Solve-RD ClinVar-based reanalysis of 1522 index cases from ERN-ITHACA reveals common pitfalls and misinterpretations in exome sequencing. *Genet Med*. 2023 Jan 20;25(4):100018.
- Structural variant calling and clinical interpretation in 6224 unsolved rare disease exomes. Demidov G, Laurie S, …, Scala M, …, Ossowski S. *Eur J Hum Genet*. 2024 Aug;32(8):998-1004.
- An interconnected data infrastructure to support large-scale rare disease research. Johansson LF, Laurie S, …; Solve-RD consortium. *Gigascience*. 2024 Jan 2;13:giae058.

- International League Against Epilepsy Consortium on Complex Epilepsies in:

- GWAS meta-analysis of over 29,000 people with epilepsy identifies 26 risk loci and subtype-specific genetic architecture. *Nat Genet*. 2023 Sep;55(9):1471-1482.

- Telethon Undiagnosed Diseases Program in:

- De novo missense variants in phosphatidylinositol kinase PIP5KIγ underlie a neurodevelopmental syndrome associated with altered phosphoinositide signaling. *Am J Hum Genet*. 2023 Aug 3;110(8):1377-1393.

- Consortium EpiPGX in:

- Testing for pharmacogenomic predictors of ppRNFL thinning in individuals exposed to vigabatrin. Boothman I, Clayton LM,…; Consortium EpiPGX; Sisodiya SM, Cavalleri GL. *Front Neurosci*. 2023 Sep 8;17:1156362.

REVIEWER ACTIVITY

Reviewer for the following journals: *iScience, Scientific Reports, Journal of Medical Genetics, Clinical Genetics, Molecular Neurobiology, The Cerebellum, Neurological Sciences, Molecular Genetics & Genomic*

EDITORIAL TASKS/COLLECTIONS

TEACHING ACTIVITY

Medicine, Epilepsy Research, Epilepsia Open, Frontiers in Pediatrics, Frontiers in Neurology, Epileptic Disorders

- Guest Editor for the *Journal of Translational Genetics and Genomics*,
- Review Editor for *Frontiers in Neurology and Frontiers in Pediatrics*

AWARDS

- Lectures in medical genetics to students in Medicine and Surgery (Pediatrics course) at the University of Genoa (Academic year 2023/2024; 2024/2025)
- Group teaching to students in Medicine and Surgery (Pediatrics course, seminars in pediatric neurology) at the University of Genoa (Academic year 2023/2024; 2024/2025)
- Lecture in the PhD course in Neuroscience at the University of Genova (Academic year 2023, Summer School, II edition) about 'Novel disease gene discovery approaches for Neuromuscular Disorders', September 11th – 15th, 2023

- '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.'
- ESHG Conference Fellowship at the European Society of Human Genetics conference (ESHG 2024, Berlin, June 1-4)

RESEARCH GRANTS

- Co-PI (2023): 'Sleep as a Window for expAnding knowledge about Neurodevelopment of very preterm infants: a neurobiological prospective study on sleep neurophysiology, genetics, and neuroendocrinology. The SWAN project.' Grant provided by the University of Genoa (Funding: 40,000 Euros)
- **PI (2024): Project 'Sequencing whole genome and RNA to unveil the genetic etiology of unsolved neuromuscular disorders'.** Grant provided by the University of Genoa (Funding: 15,000 Euros)

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