UNIVERSITÀ DEGLI STUDI DELL'AQUILA

DISCAB
Dipartimento di Scienze Cliniche Applicate e Biotecnologiche

CURRICULUM VITAE

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PERSONAL INFORMATION	Name and Surname:Vincenzo Salpietro Damiano Department: Department of Clinical Sciences and Applied Biotechnologies Address (work): Via Vetoio, Coppito City: L'Aquila, 67100 Nation:Italy E-mail address (work): **vincenzo.salpietrodamiano@univaq.it**

CURRENT POSITION	Full Professor of Paediatrics

EDUCATION OTHER QUALIFICATIONS	(Medical Degree (University of Messina, July 2009) Paediatric Residency (University of Messina & University of Pavia, Italy, May 2015) Pediatric Neurology Fellowship (Imperial College, September 2014) Master of Philosophy (University College London, November 2018)

ACADEMIC APPOINTMENTS	Honorary Lecturer of Pediatric Neurogenetics, Institute of Neurology, University College London (UCL), London, UK (December 2018 till now) Associate Professor of Paediatrics, University of Genoa and Children's Hospital "Istituto Giannina Gaslini", Genoa, Italy (December 2021) Full Professor of Paediatrics, University of L'Aquila, L'Aquila (April 2022)

CLINIC APPOINTMENTS	Head of the Outpatient clinic for "Neurodevelopmental Disorders Genomics" in Gaslini Children's Hospital, University of Genoa, Italy (2019-2021) Head of the Pediatric Outpatient and Inpatient Department at the University of L'Aquila, University Hospital "San Salvatore" (since April 2022).

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TEACHING EXPERIENCE	Mentoring medical students or Master students from University College London during their attendance in the Department of Molecular Neuroscience or in the Neurogenetics Clinic (UCLH) (since September 2015 till December 2018). Lectures of paediatric neurology and neurogenetics to medical students and residents of different specialties in University of Genoa since January 2019 Assistant Professor of Pediatrics (2018-2021) and Associate Professor of Pediatrics (since December 2021) in the University of Genoa, Italy and Children's Hospital "G. Gaslini". Full Professor of Pediatrics (since April 2022) in the University of L'Aquila Activites include mentoring and teaching of medical students, paediatrics Residents, PhD Students, and support in the preparation of scientific manuscripts or Thesis.

RESEARCH ACTIVITIES	I am a Paediatrician and a Professor of Pediatrics with a special clinical and academic interest in Paediatric Neurology and Neurogenetics. I completed my Medical studies in Italy (University of Messina) in 2009 and my training as a Paediatrician and a Paediatric Neurologist between Italy (University of Messina, University of Pavia; period: 2010-2015) and United Kingdom (Imperial College London; period: 2013-2014). Between September 2013 and September 2014, I attended the Molecular Biomedicine Lab at the Department of Medicine, Imperial College London as part of my Clinical Research Fellowship in Paediatric Neurology and Neurosciences. Since July 2015, I was appointed as Clinical Research Associate of Neurogenetics at the Department of Molecular Neuroscience in the Institute of Neurology, University College London (UCL) where I worked with Prof. Henry Houlden and Prof. James E. Rothman until December 2018. Since January 2019, I have been appointed as an University Lecturer of Paediatrics and Paediatric Neurology in the Children's Hospital "Istituto Giannina Gaslini", University of Genoa in Italy where I became Associate Professor in 2021. Also, I hold a double affiliation working part-time as Honorary Clinical senior Lecturer at the Department of Neuromuscular diseases in the Institute of Neurology, UCL, London. Since April 2022 I moved to Abruzzo in Central Italy and was appointed as a Full Professor of Pediatrics in the University of L'Aquila. My research work is mostly focused on investigating the genetic alterations causing inherited neurological disorders in children. I have identified (as Principal Investigator/First Author) mutations in the genes *VAMP1* and *VAMP2* associated with novel childhood-onset neuromuscular and central synaptopathies (Salpietro et al., Ann Neurol 2017, Salpietro et al., Am J Hum Genet 2019). I also identified a novel gene causing PEHO syndrome (Salpietro et al., Brain 2017), a novel

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	gene responsible for a phenotype of combined inherited neuropathy, polydactyly and epilepsy (Salpietro et al., Hum Mut 2017), a new gene responsible for chorea and dystonia (Salpietro et al., Mov Dis 2017). I recently identified a novel genetic neurodevelopmental disorder associated with genetically determined defects of the GluA2 subunit of the AMPA receptor, the main receptor involved in brain fast synaptic excitatory transmission (Salpietro et al., Nat Comm 2019). I also identified mutations in the novel channel KCNA6 underlying distinct epileptic phenotypes in children (Salpietro V et al., Epilepsia 2023) and mutations in the GTPBP genes in patients affected with ectodetermal neurodevelopmenta disorders (Salpietro V et al, under preparation). In a number of these newly discovered genes and phenotypes I collaborated with different labs in establishing animal models mimicking the novel human genetic diseases, using either knock-out mice (Salpietro et al., Ann Neurol. 2017) or hypomorphic Drosophila Melanogaster (fruit-flies) models (Salpietro et al., Hum Mut. 2017). Currently, I am working in elucidating the genetic and metabolic molecular basis of a number of other rare neurological disorders, including epileptic encephalopathies, childhood-onset movement disorders, familial sleep disorders and developmental synaptopathies with epilepsy and autistic features. As the result of my research work, I published during the last 5 years >100 full-length manuscripts in international peer reviewed journals.

RESPONSIBILITY IN ACADEMIC ACTIVITIES	Full Professor of Pediatrics in Italy International Coordinator of "SYNaPS Study Group" (based in London, UK)

EDITORIAL BOARD, EDITORIAL ACTIVITIES, SOCIETY MEMBERSHIP	Frontiers in Neurology (Indexed in Pubmed/Medline) Position: Associate Editor. Neuropediatrics Specialty Section (web link: http://www.frontiersin.org/Neuropediatrics/editorialboard?field=Neurology) Frontiers in Pediatrics (Indexed in Pubmed/Medline) Position: Associate Editor. Neuropediatrics Specialty Section (web link: http://www.frontiersin.org/Neuropediatrics/editorialboard?field=Neurology) Journal of Pediatric Neurology (Indexed in Scopus/Embase) Position: Associate Editor Former member of "The Genetics Society" (UK) till 2021 Current Member of the Italian Society of Pediatrics and the International Child Neurology Association

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SCIENTIFIC ACHIEVEMENTS BIBLIOMETRIC INDICATORS	**Scopus Author ID:** **http://orcid.org** (Hirsch (H) Index, i10-Hirsch (H), normalized Index, total number of quotes, median number of quotes by article

SELECTED PUBLICATIONS	**Salpietro V**, Galassi Deforie V, Efthymiou S, O'Connor E, Marcé-Grau A, Maroofian R, Striano P, Zara F, Morrow MM; SYNAPS Study Group; Reich A, Blevins A, Sala-Coromina J, Accogli A, Fortuna S, Alesandrini M, Au PYB, Singhal NS, Cogne B, Isidor B, Hanna MG, Macaya A, Kullmann DM, Houlden H, Männikkö R. De novo KCNA6 variants with attenuated KV 1.6 channel deactivation in patients with epilepsy. Epilepsia. 2023 Feb;64(2):443-455. **[First Author]** **IF 6.740** Editore: Wiley Calì E, Lin SJ, Rocca C, Sahin Y, Al Shamsi A, El Chehadeh S, Chaabouni M, Mankad K, Galanaki E, Efthymiou S, Sudhakar S, Athanasiou-Fragkouli A, Çelik T, Narlı N, Bianca S, Murphy D, De Carvalho Moreira FM; SYNaPS Study Group, Andrea Accogli, Petree C, Huang K, Monastiri K, Edizadeh M, Nardello R, Ognibene M, De Marco P, Ruggieri M, Zara F, Striano P, Şahin Y, Al-Gazali L, Abi Warde MT, Gerard B, Zifarelli G, Beetz C, Fortuna S, Soler M, Valente EM, Varshney G, Maroofian R, **Salpietro V#**, Houlden H. A homozygous MED11 C-terminal variant causes a lethal neurodegenerative disease. Genet Med. 2022 Oct;24(10):2194-2203. **[Corresponding Author]** **IF 8.864** Editore: Elsevier Mastrangelo M, Salpietro V, Sullivan J. Genetically determined epilepsies: Perspectives in the era of precision medicine. *Front Neurol* 2022 Oct 11;13:1036846. **[Second Author]** **IF 4.086** Editore: Frontiers Media SA Cuccurullo C, Miele G, Piccolo G, Bilo L, D'Amico A, Fratta M, Striano P, Lorenzo Ugga L, **Salpietro V**, Coppola A. Hydranencephaly and Seckel syndrome type 4 due to a novel *CENPJ* mutation: a case report and review of the literature. *Eur J Med Genet* 2022 Dec;65(12):104659. **IF 2.465** Editore: Elsevier Borgia P, Baldassari S, Pedemonte N, Alkhunaizi E, D'Onofrio G, Tortora D, Calì E, Scudieri P, Balagura G, Musante I, Diana MC, Pedemonte M, Vari MS, Iacomino M, Riva A, Chimenz R, Mangano GD, Mohammadi MH, Toosi

MB, Ashrafzadeh F, Imannezhad S, Karimiani EG, Accogli A, Schiaffino MC, Maghnie M, Soler MA, Echiverri K, Abrams CK, Striano P, Fortuna S, Maroofian R, Houlden H, Zara F, Fiorillo C, **Salpietro V.** Genotype-phenotype correlations and disease mechanisms in PEX13-related Zellweger spectrum disorders**.** *Orphanet J Rare Dis* 2022 Jul 19;17(1):286.
[Last Author]
IF 4.302
Editore: BMC

Accogli A, Lu S, Musante I, Scudieri P, Rosenfeld JA, Severino M, Baldassari S, Iacomino M, Riva A, Balagura G, Piccolo G, Minetti C, Roberto D, Xia F, Razak R, Lawrence E, Hussein M, E Yih-Herng Chang, Holick M, Calì E, Aliberto E, De-Sarro R, Gambardella A, Undiagnosed Diseases Network, SYNaPS Study Group, Emrick L, McCaffery PJA, Clagett-Dame M, Marcogliese PC, Bellen HJ, Lalani SR, Zara F, Striano P, **Salpietro V**. Loss of Neuron Navigator 2 Impairs Brain and Cerebellar Development
Cerebellum 2022. Feb 26. doi: 10.1007/s12311-022-01379-3. Online ahead of print.
[Last Author]
IF 3.648
Editore: Springer

Spoto G, Valentini G, Saia MC, Butera A, Amore G, **Salpietro V***, Nicotera AG, Di Rosa G. Synaptopathies in Developmental and Epileptic Encephalopathies: a focus on presynaptic dysfunction. *Front Neurol* 2022 Mar 8;13:826211
[*Corresponding Author]
IF 4.086
Editore: Frontiers Media SA

Cali E, Rocca C, **Salpietro V#**, Houlden H. Epileptic Phenotypes Associated With SNAREs and Related Synaptic Vesicle Exocytosis Machinery. *Front Neurol*. 2022 Jan 13;12:806506
[Corresponding Author]
IF 4.086
Editore: Frontiers Media SA

Koko M, Krause R, Sander T, Bobbili DR, Nothnagel M, May P, Lerche H; Epi25 Collaborative (**Salpietro V**). Distinct gene-set burden patterns underlie common generalized and focal epilepsies. *EBioMedicine*. 2021 Oct;72:103588.
IF 11.205
Editore: Elsevier

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	Casto C, Dipasquale V, Ceravolo I, Gambadauro A, Aliberto E, Galletta K, Granata F, Ceravolo G, Riva A, Piccolo G, Cutrupi MC, Di Rosa G, Gitto E, Efthymiou S, **Salpietro V#**, Houlden H, Chimenz R. Prominent and Regressive brain developmental disorders associated with Nance-Horan syndrome. *Brain Sci* 2021. *In Press*. **[Corresponding Author]** **IF 3.333** Editore: MDPI The Covid-19 Host Genetic Initiative (**Salpietro V**). Mapping the human genetic architecture of COVID-19 by worldwide meta-analysis. *Nature*. 2021 Dec;600(7889):472-477. **IF 69.504** Editore: Nature Piccolo G, D'Annunzio G, Amadori E, Riva A, Borgia P, Tortora D, Maghnie M, Minetti C, Gitto E, Fiorillo C, Zara F, Striano P, **Salpietro V**. Neuromuscular and neuroendocrinological features associated with ZC4H2-related arthrogryposis multiplex congenita in a Sicilian family. *Front Neurol* 2021 Jul 12;12:704747 **[Last Author]** **IF 4.086** Editore: Frontiers Media SA Epi25 Collaborative (**Salpietro V**). Electronic address: jm4279@cumc.columbia.edu; Epi25 Collaborative. Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. *Am J Hum Genet* 2021 Apr 28:S0002-9297(21)00140-3 **IF 11.043** Editore: Cell Press 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. Biallelic Variants in *KIF17* Associated with Microphthalmia and Coloboma Spectrum. *Int J Mol Sci*. 2021 Apr 25;22(9):4471 **[Corresponding Author]** **IF 6.208** Editore: MDPI Bourinaris T, Athanasiou A, Efthymiou S, Wiethoff S, **Salpietro V#**, Houlden H. Allelic and phenotypic heterogeneity in Junctophillin-3 related neurodevelopmental and movement disorders. *Eur J Hum Genet*. 2021 Jun;29(6):1027-1031.

	[Corresponding Author] **IF 5.351** Editore: Springer Nardello R, Mangano GD, Antona V, Fontana A, Striano P, Giorgio E, Brusco A, Mangano S, **Salpietro V**. Electroclinical features and outcome of ANKRD11-related KBG syndrome: A novel report and literature review. *Seizure*. 2021;85:151-154. **[Last Author]** **IF 3.414** Editore: Elsevier Baldassari S, Musante I, Iacomino M, Zara F, **Salpietro V#**, Scudieri P. Brain Organoids as Model Systems for Genetic Neurodevelopmental Disorders. *Front Cell Dev Biol* 2020 Oct 12;8:590119 **[Corresponding Author]** **IF 6.081** Editore: Frontiers Media SA Nardello R, Mangano GD, Fontana A, Gagliardo C, Midiri F, Borgia P, Brighina F, Raieli V, Mangano S, **Salpietro V**. Broad neurodevelopmental features and cortical anomalies associated with a novel de novo KMT2A variant in Wiedemann-Steiner syndrome. *Eur J Med Genet* 2021 Feb;64(2):104133. **[Last Author]** **IF 2.465** Editore: Elsevier Nardello R, Mangano GD, Miceli F, Fontana A, Piro E, **Salpietro V**. Benign familial infantile epilepsy associated with KCNQ3 mutation: a rare occurrence or an underestimated event? *Epileptic Disord* 2020;22(6):807-810 **[Last Author]** **IF 2.333** Editore: John Libbey Iacomino M, Baldassari S, Tochigi Y, Kosla K, Buffelli F, Torella AL, Severino MS, Paladini D, Mandarà L, Riva L, Scala M, Balagura G, Accogli A, Nigro V, Minetti C, Fulcheri E, Zara F, Bednarek AK, Striano P, Suzuki P, **Salpietro V**. Loss of Wwox perturbs neuronal migration and impairs early cortical development. *Front Neurosci* 2020 Jun 11;14:644 **[Last Author]** **IF 5.152** Editore: Frontiers Media SA

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	K, Vitobello A, Tran Mau-Them F, Faivre L, Martinez-Azorin F, Prada CE, Macaya A, Kullmann DM, Rothman JE, Krishnakumar SS, Houlden H. Mutations in the neuronal vesicular SNARE *VAMP2* affect synaptic membrane fusion and impair human neurodevelopment. *Am J Hum Genet* 2019 Apr 4;104(4):721-730. **[First Author]** **IF 11.043** Editore: Cell Press **Salpietro V**, Perez-Dueñas B, Nakashima K, San Antonio MV, Manole A, Efthymiou S, Bettencourt C, Mencacci NE, Klein C, Kelly MP, Davies CH, Kimura H, Macaya A, Houlden H. A homozygous loss-of-function mutation in PDE2A associated to early-onset hereditary chorea. *Mov Dis* 2018 Mar;33(3):482-488 **[First Author]** **IF 9.698** Editore: Wiley **Salpietro V**, Efthymiou S, Manole A, Maurya B, Wiethoff S, Ashokkumar B, Cutrupi MC, Dipasquale V, Manti S, Botia JA, Vandrovcova J, Bettencourt C, Mankad K, Mukherjee A, Mutsuddi M, Houlden H. A loss-of-function homozygous mutation in DDX59 implicates a conserved RNA helicase in nervous system development and function. *Hum Mut*. 2018 Feb;39(2):187-192 **[First Author]** **IF 4.700** Editore: Wiley **Salpietro V**, Zollo M, Vandrovcova J, Ryten M, Botia JA, Ferrucci V, Efthymiou S, Al Mutairi F, Bertini E, Tartaglia M, SYNAPS Study Group, Houlden H. The phenotypic and molecular spectrum of PEHO syndrome and PEHO-like disorders. *Brain* 2017 Aug 1;140(8):e49. **[First Author]** **IF 15.255** **Editore: Oxford University Press** **Salpietro V**, Lin W, Vedove AD, Storbeck M, Liu Y, Efthymiou S, Manole A, Wiethoff S, Ye Q, Saggar A, McElreavey K, Krishnakumar SS, SYNAPS Study Group, Pitt M, Bello OD, Rothman JE, Basel-Vanagaite L, Hubshman MW, Aharoni S, Manzur AY, Wirth B, Houlden H. Homozygous mutations in VAMP1 cause a presynaptic congenital myasthenic syndrome. *Ann Neurol* 2017 Apr;81(4):597-603 **[First Author]** **IF 11.274** Editore: Wiley

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L'AQUILA, 04/04/23