AI FINI DELLA PUBBLICAZIONE

Decreto Rettore Università di Roma "La Sapienza" n 2267/2021 del 09.08.2021

SERENA GALOSI
Curriculum Vitae

Part I – GENERAL INFORMATION

Full Name	Serena Galosi
Spoken Languages	Italian, English

Part II – EDUCATION

Type	Year/s	Institution	Notes (Degree, Experience,…)
University graduation	2003-2009	Sapienza University, Rome	110/110 *summa cum laude*
Post-graduate studies: Residency	2010-2015	Sapienza University, Rome	Residency in Child Neurology and Psychiatry
PhD	2015-2019	Sapienza University, Rome	Dept. of Human Neuroscience, PhD Course in Clinical and Experimental Neurosciences
Research Scholar	2017-2018	University of California San Diego (UCSD) and Rady Children's Hospital	Research Fellowship at UCSD Dept. of Neuroscience, and Pediatric Movement Disorder Clinic and Institute for Genomic Medicine at Rady Children's Hospital

Part III – APPOINTMENTS
IIIA-Academic Appointments

Start	End	Institution	Position
July 2017	August 2018	University of California San Diego and Rady Children Hospital, Pediatric and Adult Movement Disorders Clinic, Institute for Genomic Medicine	Research Scholar

IIIB-Other Appointments: Professional Experience

Start	End	Institution	Position
April 2020	Today	Policlinico Umberto I, University Hospital (Sapienza, University of Rome), via dei Sabelli 108, 00181	Medical director in Child and Adolescent Neurology and Psychiatry (Permanent full-time employment)

		Rome	
April 2017	March 2020	Policlinico Umberto I, University Hospital (Sapienza, University of Rome), via dei Sabelli 108, 00181 Rome	Freelance employment as consultant in Child and Adolescent Neurology and Psychiatry
Sept. 2015	Sept 2016	Policlinico Umberto I, University Hospital (Sapienza, University of Rome), via dei Sabelli 108, 00181 Rome	Freelance employment as consultant in Child and Adolescent Neurology and Psychiatry
July 2016	March 2017	Assohandicap, rehabilitation centre accredited and affiliated with the national health system, Via Pietro Nenni 16, Marino (RM)	Freelance employment as consultant in Child and Adolescent Neurology and Psychiatry

Part IV – TEACHING EXPERIENCE

Year	Institution	Lecture/Course
2016/ 2017	Sapienza University, Rome	Advanced training course in Pediatric Movement Disorders [Disturbi del Movimento nel bambino nuove acquisizioni scientifiche e Strategie diagnostiche, 29181]
2017/ 2018	Sapienza University, Rome	Advanced training course in Pediatric Neurology [Neurologia Infantile: nuove acquisizioni scientifiche, nuove malattie, nuove strategie diagnostiche, nuovi approcci terapeutici, 29807]
2019/ 2020	Sapienza University, Rome	Occupational Therapy Program, Degree Code: L/SNT2, Course: *General Basis of occupational therapy in developmental age, Principles of Child Neurology and Psychiatry* (20 hours, 1 CFU)
2020/ 2021	Sapienza University, Rome	Occupational Therapy Program, Degree Code: L/SNT2, Course: *General Basis of occupational therapy in developmental age*, *Principles of Child Neurology and Psychiatry* (20 hours, 1 CFU)
2019/ 2020	Sapienza University, Rome	Faculty of Pharmacy and Medicine/Faculty Medicine and Dentistry, Degree Code: LM-41, Master A, Course: *Pediatrics*, Series of lectures on *Neurodevelopmental disorders, Pediatric Movement Disorders, and diagnosis of rare genetic neurological diseases* (13 hours)
2020/ 2021	Sapienza University, Rome	Faculty of Pharmacy and Medicine/Faculty Medicine and Dentistry, Degree Code: LM-41, Master A, Course: *Pediatrics*, Series of lectures on *Neurodevelopmental disorders,*

		Pediatric Movement Disorders, and diagnosis of rare genetic neurological diseases (13 hours)
2021	Sapienza University, Rome	Master in Rare diseases, Director Sandra Giustini, Lecture on *Rare Pediatric Movement Disorders of genetic etiology*
2021	Alma Mater Studiorum Università di Bologna	Master in inborn metabolic diseases and Newborn Screening, Lecture on *Primary and secondary Neurotransmitter Disorders*

Part V - SOCIETY MEMBERSHIPS, AWARDS AND HONORS

Year	Title
2016-2021	Member of the International Parkinson and Movement Disorder Society (MDS)
2019-2021	SINPIA Neurology Group, Member of the Scientific Committee, Workgroup on Pediatric Movement Disorders
2018	**Travel grant** for the participation to the International Congress of The International Parkinson and Movement Disorder Society (MDS) (5-9 October 2018, Hong Kong) associated with the presentation of the following posters: ▪ Galosi, S., Pons, M. R., Zouvelou, B., Leuzzi, V., & Friedman, J. (2018, October). High amplitude jerky tremor in developmental dopamine deficiency. In *MOVEMENT DISORDERS* (Vol. 33, pp. S212-S213). 111 RIVER ST, HOBOKEN 07030-5774, NJ USA: WILEY. ▪ Galosi, S., Schirinzi, T., Bertini, E., Haas, R., Santorelli, F. M., Leuzzi, V., & Friedman, J. R. (2018, October). Dystonia ataxia (DYTCA) syndrome with prominent handwriting deterioration associated with ADCK3 mutation: two new cases and an overview of the literature. In *MOVEMENT DISORDERS* (Vol. 33, pp. S226-S227). 111 RIVER ST, HOBOKEN 07030-5774, NJ USA: WILEY.
2015	Award for Best oral presentation; VI Congresso nazionale Società Italiana Malattie Metaboliche e screening neonatale (SIMMESN) (16-18 December, Florence)

Part VI - FUNDING INFORMATION [GRANTS AS PI-PRINCIPAL INVESTIGATOR OR I-INVESTIGATOR]

Year	Title	Program	Grant value
2021	Movement Disorders 0-18: a research platform integrating dataset for gene identification and disease-specific register	Bando Fondazione Mariani 2020 Neurologia Infantile (Included as Collaborator)	80.000 £

Part VII – RESEARCH ACTIVITIES

Keywords	Brief Description
Pediatric Movement disorders	Clinical, biochemical, and genetic characterization of rare and new genetic disorders presenting with movement disorders during childhood including hyperkinetic (dystonia, chorea, myoclonus), hypokinetic (parkinsonism), and paroxysmal motor disorders. Special interest on the following recently described genes: *GNAO1*, *KMT2B*, *DHDDS*
Neurometabolic disorders	Metabolic movement disorders presenting during childhood: early clinical and biochemical markers for diagnosis, genetic characterization and outcome. Special interest in Primary and secondary monoamine disorders, Congenital Disorders of Glycosylation, lipid metabolism and mitochondrial disorders (Primary CoQ10 Deficiency, WARS2 deficiency).
Precision Medicine/ disease modeling	Functional characterization of identified genetic variants and disease modeling in multiple systems (*c.elegans*/yeast/cells including generation of iPSCs, mice) of the following genes, both as past and ongoing research projects: *WARS2*, *GNAO1*, *KMT2B*, and *DHDDS*. For *GNAO1* and *DHDDS* functional studies on mice and cellular models are ongoing to dissect the pathophysiology and design tailored therapeutic approaches. Collaborations for functional studies: Department of Oncology and Molecular Medicine, Istituto Superiore di Sanità; Vascular Biology and Therapeutics Program, Yale University School of Medicine, New Haven, CT 06520, USA; Department of Pharmacology, Yale University School of Medicine, New Haven, CT 06520, USA; Department of Physiology and Pharmacology, Sapienza University of Rome, Rome, Italy; Genetics and Rare Diseases Research Division, Ospedale Pediatrico Bambino Gesù, IRCCS, Rome, Italy.

Part VIIA-Publications

1. Galatolo D, De Michele G, Silvestri G, Leuzzi V, Casali C, Musumeci O, Antenora A, Astrea G, Barghigiani M, Battini R, Battisti C, Caputi C, Cioffi E, De Michele G, Dotti MT, Fico T, Fiorillo C, **Galosi S**, Lieto M, Malandrini A, Melone MAB, Mignarri A, Natale G, Pegoraro E, Petrucci A, Ricca I, Riso V, Rossi S, Rubegni A, Scarlatti A, Tinelli F, Trovato R, Tedeschi G, Tessa A, Filla A, Santorelli FM. NGS in Hereditary Ataxia: When Rare Becomes Frequent. Int J Mol Sci. 2021 Aug 6;22(16):8490. doi: 10.3390/ijms22168490. PMID: 34445196; PMCID: PMC8395181. IF 5.923

2. **Galosi S**, Edani BH, Martinelli S, Hansikova H, Eklund EA, Caputi C, Masuelli L, Corsten-Janssen N, Srour M, Oegema R, Bosch DGM, Ellis CA, Amlie-Wolf L, Accogli A, Atallah I, Averdunk L, Barañano KW, Bei R, Bagnasco I, Brusco A, Demarest S, Alaix AS, Di Bonaventura C, Distelmaier F, Elmslie F, Gan-Or Z, Good JM, Gripp K, Kamsteeg EJ, Macnamara E, Marcelis C, Mercier N, Peeden J, Pizzi S, Pannone L, Shinawi M, Toro C, Verbeek NE, Venkateswaran S, Wheeler PG, Zdrazilova L, Zhang R, Zorzi G, Guerrini R,

Sessa WC, Lefeber D, Tartaglia M, Hamdan FF, Grabińska KA, Leuzzi V. De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. Brain. 2021 Aug 11:awab299. doi: 10.1093/brain/awab299. Epub ahead of print. PMID: 34382076. IF 13.501

3. Ciolfi A, Foroutan A, Capuano A, Pedace L, Travaglini L, Pizzi S, Andreani M, Miele E, Invernizzi F, Reale C, Panteghini C, Iascone M, Niceta M, Gavrilova RH, Schultz-Rogers L, Agolini E, Bedeschi MF, Prontera P, Garibaldi M, **Galosi S**, Leuzzi V, Soliveri P, Olson RJ, Zorzi GS, Garavaglia BM, Tartaglia M, Sadikovic B. Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. Clin Epigenetics. 2021 Aug 11;13(1):157. doi: 10.1186/s13148-021-01145-y. PMID: 34380541; PMCID: PMC8359374. IF 5.028

4. De Michele G, Galatolo D, **Galosi S**, Mignarri A, Silvestri G, Casali C, Leuzzi V, Ricca I, Barghigiani M, Tessa A, Cioffi E, Caputi C, Riso V, Dotti MT, Saccà F, De Michele G, Cocozza S, Filla A, Santorelli FM. Episodic ataxia and severe infantile phenotype in spinocerebellar ataxia type 14: expansion of the phenotype and novel mutations. J Neurol. 2021 Jul 22. doi: 10.1007/s00415-021-10712-5. Epub ahead of print. PMID: 34292398. IF 4.849

5. Leone C, **Galosi S**, Mollica C, Fortunato M, Possidente C, Milone V, Misuraca S, Berillo L, Truini A, Cruccu G, Ferrara M, Terrinoni A. Dissecting pain processing in adolescents with Non-Suicidal Self Injury: Could suicide risk lurk among the electrodes? Eur J Pain. 2021 Sep;25(8):1815-1828. doi: 10.1002/ejp.1793. Epub 2021 May 31. PMID: 33982830. IF 3.931

6. Riso V, Galatolo D, Barghigiani M, **Galosi S**, Tessa A, Ricca I, Rossi S, Caputi C, Cioffi E, Leuzzi V, Casali C, Santorelli FM, Silvestri G. A next generation sequencing-based analysis of a large cohort of ataxic patients refines the clinical spectrum associated with spinocerebellar ataxia 21. Eur J Neurol. 2021 Aug;28(8):2784-2788. doi: 10.1111/ene.14868. Epub 2021 May 27. PMID: 33851480; PMCID: PMC8361921. IF 6.089

7. **Galosi S**, Martinelli S, Pannone L, Terrinoni A, Venditti M, Pizzi S, Ciolfi A, Chillemi G, Gigliotti F, Cesario S, Tartaglia M, Leuzzi V. Co-occurring SYNJ1 and SHANK3 variants in a girl with intellectual disability, early-onset parkinsonism and catatonic episodes. Parkinsonism Relat Disord. 2021 Mar;84:5-7. doi: 10.1016/j.parkreldis.2020.12.022. Epub 2021 Jan 12. PMID: 33515856. IF 4.891

8. Cif L, Demailly D, Lin JP, Barwick KE, Sa M, Abela L, Malhotra S, Chong WK, Steel D, Sanchis-Juan A, Ngoh A, Trump N, Meyer E, Vasques X, Rankin J, Allain MW, Applegate CD, Attaripour Isfahani S, Baleine J, Balint B, Bassetti JA, Baple EL, Bhatia KP, Blanchet C, Burglen L, Cambonie G, Seng EC, Bastaraud SC, Cyprien F, Coubes C, d'Hardemare V; Deciphering Developmental Disorders Study, Doja A, Dorison N, Doummar D, Dy-Hollins ME, Farrelly E, Fitzpatrick DR, Fearon C, Fieg EL, Fogel BL, Forman EB, Fox RG; Genomics England Research Consortium, Gahl WA, **Galosi S**, Gonzalez V, Graves TD, Gregory A, Hallett M, Hasegawa H, Hayflick SJ, Hamosh A, Hully M, Jansen S, Jeong SY, Krier JB, Krystal S, Kumar KR, Laurencin C, Lee H, Lesca G, François LL, Lynch T, Mahant N, Martinez-Agosto JA, Milesi C, Mills KA, Mondain M, Morales-Briceno H; NIHR BioResource, Ostergaard JR, Pal S, Pallais JC, Pavillard F, Perrigault PF, Petersen AK, Polo G, Poulen G, Rinne T, Roujeau T, Rogers C, Roubertie A, Sahagian M, Schaefer E, Selim L,

Selway R, Sharma N, Signer R, Soldatos AG, Stevenson DA, Stewart F, Tchan M; Undiagnosed Diseases Network, Verma IC, de Vries BBA, Wilson JL, Wong DA, Zaitoun R, Zhen D, Znaczko A, Dale RC, de Gusmão CM, Friedman J, Fung VSC, King MD, Mohammad SS, Rohena L, Waugh JL, Toro C, Raymond FL, Topf M, Coubes P, Gorman KM, Kurian MA. KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. Brain. 2020 Dec 5;143(11):3242-3261. doi: 10.1093/brain/awaa304. PMID: 33150406; PMCID: PMC7719027. IF 13.501

9. Leuzzi V, Nardecchia F, Pons R, **Galosi S**. Parkinsonism in children: Clinical classification and etiological spectrum. Parkinsonism Relat Disord. 2021 Jan;82:150-157. doi: 10.1016/j.parkreldis.2020.10.002. Epub 2020 Oct 21. PMID: 33109474. IF 4.801

10. Pollini L, **Galosi S**, Tolve M, Caputi C, Carducci C, Angeloni A, Leuzzi V. *KCND3*-Related Neurological Disorders: From Old to Emerging Clinical Phenotypes. Int J Mol Sci. 2020 Aug 13;21(16):5802. doi: 10.3390/ijms21165802. PMID: 32823520; PMCID: PMC7461103. IF 4.556

11. Marti-Sanchez L, Baide-Mairena H, Marcé-Grau A, Pons R, Skouma A, López-Laso E, Sigatullina M, Rizzo C, Semeraro M, Martinelli D, Carrozzo R, Dionisi-Vici C, González-Gutiérrez-Solana L, Correa-Vela M, Ortigoza-Escobar JD, Sánchez-Montañez Á, Vazquez É, Delgado I, Aguilera-Albesa S, Yoldi ME, Ribes A, Tort F, Pollini L, **Galosi S**, Leuzzi V, Tolve M, Pérez-Gay L, Aldamiz-Echevarría L, Del Toro M, Arranz A, Roelens F, Urreizti R, Artuch R, Macaya A, Pérez-Dueñas B. Delineating the neurological phenotype in children with defects in the ECHS1 or HIBCH gene. J Inherit Metab Dis. 2021 Mar;44(2):401-414. doi: 10.1002/jimd.12288. Epub 2020 Aug 16. PMID: 32677093. IF 4.982

12. Ng A, **Galosi S**, Salz L, Wong T, Schwager C, Amudhavalli S, Gelineau-Morel R, Chowdhury S; Rady Children's Institute for Genomic Medicine Investigators, Friedman J. Failure to thrive - an overlooked manifestation of KMT2B-related dystonia: a case presentation. BMC Neurol. 2020 Jun 16;20(1):246. doi: 10.1186/s12883-020-01798-x. PMID: 32546208; PMCID: PMC7296679. IF 2.29

13. Martinelli S, Cordeddu V, **Galosi S**, Lanzo A, Palma E, Pannone L, Ciolfi A, Di Nottia M, Rizza T, Bocchinfuso G, Traversa A, Caputo V, Farrotti A, Carducci C, Bernardini L, Cogo S, Paglione M, Venditti M, Bentivoglio A, Ng J, Kurian MA, Civiero L, Greggio E, Stella L, Trettel F, Sciaccaluga M, Roseti C, Carrozzo R, Fucile S, Limatola C, Di Schiavi E, Tartaglia M, Leuzzi V. Co-occurring WARS2 and CHRNA6 mutations in a child with a severe form of infantile parkinsonism. Parkinsonism Relat Disord. 2020 Mar;72:75-79. doi: 10.1016/j.parkreldis.2020.02.003. Epub 2020 Feb 15. PMID: 32120303. IF 3.926

14. Pollini L, Tolve M, Nardecchia F, **Galosi S**, Carducci C, di Carlo E, Carducci C, Leuzzi V. Multiple sclerosis and intracellular cobalamin defect (*MMACHC*/*PRDX1*) comorbidity in a young male. Mol Genet Metab Rep. 2020 Jan 7;22:100560. doi: 10.1016/j.ymgmr.2019.100560. PMID: 32099815; PMCID: PMC7026611. IF 1.83

15. **Galosi S**, Nardecchia F, Leuzzi V. Treatable Inherited Movement Disorders in Children: Spotlight on Clinical and Biochemical Features. Mov Disord Clin Pract. 2020 Feb 4;7(2):154-166. doi: 10.1002/mdc3.12897. PMID: 32071932; PMCID: PMC7011670.

16. Pollini L, **Galosi S**, Nardecchia F, Musacchia F, Castello R, Nigro V, Leuzzi V. Parkinsonism, Intellectual Disability, and Catatonia in a Young Male With *MECP2* Variant. Mov Disord Clin Pract. 2019 Nov 21;7(1):118-119. doi: 10.1002/mdc3.12865. PMID: 31970230; PMCID: PMC6962672.

17. **Galosi S**, Barca E, Carrozzo R, Schirinzi T, Quinzii CM, Lieto M, Vasco G, Zanni G, Di Nottia M, Galatolo D, Filla A, Bertini E, Santorelli FM, Leuzzi V, Haas R, Hirano M, Friedman J. Dystonia-Ataxia with early handwriting deterioration in COQ8A mutation carriers: A case series and literature review. Parkinsonism Relat Disord. 2019 Nov;68:8-16. doi: 10.1016/j.parkreldis.2019.09.015. Epub 2019 Sep 28. PMID: 31621627. IF 3.926

18. Somma A, Fossati A, Ferrara M, Fantini F, **Galosi S**, Krueger RF, Markon KE, Terrinoni A. DSM-5 personality domains as correlates of non-suicidal self-injury severity in an Italian sample of adolescent inpatients with self-destructive behaviour. Personal Ment Health. 2019 Nov;13(4):205-214. doi: 10.1002/pmh.1462. Epub 2019 Jul 28. PMID: 31353830. IF 1.929

19. Carecchio M, Invernizzi F, Gonzàlez-Latapi P, Panteghini C, Zorzi G, Romito L, Leuzzi V, **Galosi S**, Reale C, Zibordi F, Joseph AP, Topf M, Piano C, Bentivoglio AR, Girotti F, Morana P, Morana B, Kurian MA, Garavaglia B, Mencacci NE, Lubbe SJ, Nardocci N. Frequency and phenotypic spectrum of KMT2B dystonia in childhood: A single-center cohort study. Mov Disord. 2019 Oct;34(10):1516-1527. doi: 10.1002/mds.27771. Epub 2019 Jun 19. PMID: 31216378. IF 8.679

20. Caputi C, Tolve M, **Galosi S**, Inghilleri M, Carducci C, Angeloni A, Leuzzi V. PNKP deficiency mimicking a benign hereditary chorea: The misleading presentation of a neurodegenerative disorder. Parkinsonism Relat Disord. 2019 Jul;64:342-345. doi: 10.1016/j.parkreldis.2019.03.012. Epub 2019 Apr 1. PMID: 30956058. IF 3.926

21. Schirinzi T, Garone G, Travaglini L, Vasco G, **Galosi S**, Rios L, Castiglioni C, Barassi C, Battaglia D, Gambardella ML, Cantonetti L, Graziola F, Marras CE, Castelli E, Bertini E, Capuano A, Leuzzi V. Phenomenology and clinical course of movement disorder in GNAO1 variants: Results from an analytical review. Parkinsonism Relat Disord. 2019 Apr;61:19-25. doi: 10.1016/j.parkreldis.2018.11.019. Epub 2018 Nov 16. PMID: 30642806. IF 3.926

22. Zima L, Ceulemans S, Reiner G, **Galosi S**, Chen D, Sahagian M, Haas RH, Hyland K, Friedman J. Paroxysmal motor disorders: expanding phenotypes lead to coalescing genotypes. Ann Clin Transl Neurol. 2018 Jul 17;5(8):996-1010. doi: 10.1002/acn3.597. PMID: 30128325; PMCID: PMC6093839. IF 4.7

23. Danti FR, **Galosi S**, Romani M, Montomoli M, Carss KJ, Raymond FL, Parrini E, Bianchini C, McShane T, Dale RC, Mohammad SS, Shah U, Mahant N, Ng J, McTague A, Samanta R, Vadlamani G, Valente EM, Leuzzi V, Kurian MA, Guerrini R. *GNAO1* encephalopathy: Broadening the phenotype and evaluating treatment and outcome. Neurol Genet. 2017 Mar 21;3(2):e143. doi: 10.1212/NXG.0000000000000143. PMID: 28357411; PMCID: PMC5362187. IF 2.74

24. Laghi F, Terrinoni A, Cerutti R, Fantini F, **Galosi S**, Ferrara M, Bosco FM. Theory of mind in non-suicidal self-injury (NSSI) adolescents. Conscious Cogn. 2016 Jul;43:38-47. doi: 10.1016/j.concog.2016.05.004. Epub 2016 May 26. PMID: 27236355. IF 2.584

25. Mastrangelo M, Caputi C, **Galosi S**, Giannini MT, Leuzzi V. Transdermal rotigotine in the treatment of aromatic L-amino acid decarboxylase deficiency. Mov Disord. 2013 Apr;28(4):556-7. doi: 10.1002/mds.25303. Epub 2013 Feb 6. PMID: 23390030. IF 6.773

Part VIIB-Books

1. Manuale di neuropsichiatria infantile e dell'adolescenza, Casa Editrice SEU, 2018- Cp 66: Segni e sintomi associati a malattie metaboliche ereditarie in neuropsichiatria infantile. Autori: Vincenzo Leuzzi, Serena Galosi

Part VIIC- Invited speaker at International or National Congresses

2021	International Parkinson and Movement Disorder Society (MDS) International Congress	*Metabolic Movement Disorders presenting during childhood*
2020	International Parkinson and Movement Disorder Society (MDS) International Congress	*Pediatric Hyperkinetic Movement Disorders: Approach to a Child Who Moves Too Much*
2020	GNAO1 European Conference, hosted by Famiglie GNAO1	*GNAO1 related disorders: update on clinical spectrum*
2018	Patrocinio SINP e SINPIA, IRCCS Istituto delle Scienze Neurologiche di Bologna (ISNB)	*Le malattie neurologiche rare e del Neurosviluppo ad esordio nei primi anni di vita, Lecture: Forme cliniche da difetto del gene GNAO1*

Part VIID- Selected oral communication at International or National Congresses

2021	14th International Congress of Inborn Errors of Metabolism (ICIEM) (21-23 November 2021, Sydney)	*De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder in the spectrum of myoclonus syndromes intersecting glycosylation and organelle disorders*
2021	International Parkinson and Movement Disorder Society (MDS) International Congress (virtual)	*MDS Videochallenge, case presentation*
2020	Pediatric Movement Disorder Congress, Barcelona	*KGD4 biallelic variants in two siblings with bilateral striatal necrosis: a new gene of Krebs cycle associated with Leigh syndrome*
2019	SSIEM Annual Symposium (2-6 September, Rotterdam)	*KGD4 biallelic variants in two siblings with bilateral striatal necrosis: a new gene of Krebs cycle associated with Leigh syndrome*

2016	Pediatric Movement Disorder Congress, Barcelona	*Clinical outcome in early-treated Sepiapterine reductase deficiency*
2015	European Metabolic Group (EMG) Conference, Venezia	*A new case of infantile parkinsonism-dystonia due nicotine receptor alteration*
2015	VI Congresso nazionale Società Italiana Malattie Metaboliche e screening neonatali (SIMMESN), Florence	*A new form of infantile parkinsonism-dystonia due nicotine receptor alteration* (Awarded as best oral communication)

Part VIIE-Editorial Experience

Peer Reviewer: Review of 6 papers published in international journals (*Brain*, *Annals of Clinical and Translational Neurology*, *Movement Disorder Clinical Practice*)

Part VIIE-Clinical Trial Experience

July 2011 – May 2015 Merck Serono Protocoll EMR700773-003: "A Phase IIIb, Multicentre, Open-Label, Randomized, Controlled Study of the Efficacy, Safety, and Population Phamacokinetics of Sapropterin Dihydrochloride (Kuvan) in Phenylketonuria (PKU) Patients <4 Years Old (SPARK)"

April 2011 – May 2014 Observational Post-Authorization Safety Study (phase IV), EMR 700773-001, Kuvan® Adult Maternal Pediatric European Registry (KAMPER)

Part VIII – Summary of Scientific Achievements

Product type	Number	Data Base	Start	End
Papers [international]	25	Pubmed	2013	2021
Papers [national]	0			
Books [scientific]	0			
Books [teaching]	1			

Total Impact factor	115,354
Total Citations	161 (Scopus)
Average Citations per Product	6.7
Hirsch (H) index	7 (Scopus)
Normalized H index*	0,63

*H index divided by the academic seniority.

Part IX– List of Selected Publications

1. **Galosi S**, Edani BH, Martinelli S, Hansikova H, Eklund EA, Caputi C, Masuelli L, Corsten-Janssen N, Srour M, Oegema R, Bosch DGM, Ellis CA, Amlie-Wolf L, Accogli A, Atallah I, Averdunk L, Barañano KW, Bei R, Bagnasco I, Brusco A, Demarest S, Alaix AS, Di

Bonaventura C, Distelmaier F, Elmslie F, Gan-Or Z, Good JM, Gripp K, Kamsteeg EJ, Macnamara E, Marcelis C, Mercier N, Peeden J, Pizzi S, Pannone L, Shinawi M, Toro C, Verbeek NE, Venkateswaran S, Wheeler PG, Zdrazilova L, Zhang R, Zorzi G, Guerrini R, Sessa WC, Lefeber D, Tartaglia M, Hamdan FF, Grabińska KA, Leuzzi V. De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. Brain. 2021 Aug 11:awab299. doi: 10.1093/brain/awab299. Epub ahead of print. PMID: 34382076. IF: 13.501; Citations:0

2. **Galosi S**, Barca E, Carrozzo R, Schirinzi T, Quinzii CM, Lieto M, Vasco G, Zanni G, Di Nottia M, Galatolo D, Filla A, Bertini E, Santorelli FM, Leuzzi V, Haas R, Hirano M, Friedman J. Dystonia-Ataxia with early handwriting deterioration in COQ8A mutation carriers: A case series and literature review. Parkinsonism Relat Disord. 2019 Nov;68:8-16. doi: 10.1016/j.parkreldis.2019.09.015. Epub 2019 Sep 28. PMID: 31621627. IF: 4.891; Citations:7

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