Curriculum Vitae

Name: BARONE Rita Maria Elisa
Citizenship: Italian
Affiliation: University of Catania, Department of Clinical and Experimental Medicine – Section of Pediatrics and Child Neuropsychiatry - Via S. Sofia 84;95123 Catania, Italy

Present Position
Professor of Child Neurology and Psychiatry - School of Medicine - Department of Clinical and Experimental Medicine, University of Catania.
Head of the Orthoptics and Assistance in Ophthalmology School, University of Catania.
Sub-Representative European Reference Network Hereditary Metabolic Disorders (MetabERN) for the Referral Center of Inborn Errors of Metabolism-Policlinico, University of Catania
Associate Professor at Consiglio Nazionale delle Ricerche (CNR) – IPCB – Catania
National Scientific Habilitation to full professor of Child Neuropsychiatry (SSD MED/39) (12/11/2020 - 12/11/2029)

Research performances
ORCiD number: 0000-0001-6302-2686
Scopus Author ID: 7006729370
ResearcherID: K-1412-2016
Bibliometric indicators (Scopus, April 2022): Number of publications 143, H-index 29; citations 2861.
https://sciprofiles.com/profile/346245

Education
1990: M.D. degree with first class honour at the medical school of the University of Catania (Italy)
1996: Specialization degree with first class honour in Child Neuropsychiatry,University of Catania (Italy)
2000: Ph.D. in Clinical and Biology of lymphoid neoplasms in childhood, University of Catania (Italy)

Clinical and Research Training:
1993 Research Fellow University of Barcelona (Spain) – Institute de Bioquimica Clinica (Prof. T. Pampols)
1995 Research Fellow University of Mainz (Germany) – Department of Pediatrics - (Prof. Michael Beck)(training activity in medical genetics and enzyme replacement therapy)
1996 Research Fellow University of Leuven (Belgium) – Department of Pediatrics (Prof. Jaak Jaeken) (training activity in molecular genetics, glycome analyses, congenital disorders of glycosylation)

Past positions
2001-2002 Research Grant at the Institute of Neurological Sciences – National Council of Research – Cosenza Programme: “Genetics of neurological childhood disorders”.
2005-2008 Research Grant at the Institute of Chemistry – National Council of Research – Catania Programme: “Glycomics of neurological childhood disorders”.
2009 Research Grant at the Neurology Clinic University of Catania:Programme: “Metabolic diseases of the central nervous systems. Clinical and proteomic characterization”.

Academic experiences
Associate Professor of Child Neurology and Psychiatry, School of Medicine, University of Catania
-University courses: Medicine and Surgery (2011 to date); Orthotic and Assistance Oftalmology (2014 to date); Occupational Therapy (2019 to date); Physiotherapy (2011-2013); Methods in Neuropsychiatric

rehabilitation techniques (2011-2013).
-School of Specializations: Child Neuropsychiatry; Physical Medicine and Rehabilitation ; Neurology; Pediatrics.
-PhD courses: International Doctorate in Neuroscience ; International Doctorate in Complex Systems for Physical, Socio-economic and Life Sciences - University of Catania

Awards and Honors
Elected member of the Steering Committee of the Italian Society for the Study of the Inherited Metabolic Diseases and Newborn Screening (2018-2021)
Member of the Advisor Board Pediatric Neurology Section – Italian National Society of Child Neuropsychiatry
Reviewer activity: REPRISE (Register of Expert Peer Reviewers for Italian Scientific Evaluation)
First Prize "Salvatore Barberi" for the best paper on Preventive Pediatric Medicine.
Organization of the II International Euroglycanet Congress on Congenital Disorders of Glycosylation, University of Catania - Catania, Italy. 3-6 Aprile, 2003
Organization of the First Orphan Europe Academy Focus Course on "Protein Glycosylation in Health and Disease" University of Catania,Italy. 1-2 April, 2003. CME accreditation.

INVITED REVIEWER
Journal of Pediatrics, Journal of Inherited Metabolic Diseases (since 2008), Clinical Genetics, Clinica Chimica Acta, European Journal of Pediatrics, Italian Journal of Pediatrics, European Journal of Human Genetics, Annals of Neurology,Pediatrics, Journal of Child Neurology, Journal of Autism and Developmental Disorders, EMBO Molecular Medicine, Trends in Molecular Medicine, Therapeutic Advances in Rare Diseases, Frontiers in Psychiatry, Frontiers in Neurology, Frontiers in Pediatrics.

Main domain of research
Since 1994 main domains of research are inherited metabolic diseases with special regard to assessment and diagnosis of neurometabolic disorders, especially lysosomal storage disorders and congenital disorders of glycosylation.

Main Scientific Achievements

1996 Participation to mapping of the locus of CDG-I (PMM2-CDG) (Genomics. 1996 Aug 1;35(3):597-9.)
1997 Participation to the Identification of the phosphomannomutase deficiency as main cause of CDG-I (J Inherit Metab Dis. 1997 Jul;20(3):447-9)
1999 Participation to the CDG nomenclature assignation international workshop (Glyconj. J. 1999; 16: 669-671)
2005 Characterization of secondary glycosylation defects in Galactosemia as dual CDG (Glycobiology. 2005 Dec;15(12):1268-76.)
2006 Participation to the first clinical trial with SRT in infantile Tay Sachs disease (Neurology. 2006 24;66(2):278-80.)
2012 Participation to Identification of the DPM2 gene (DPM2-CDG) (Annals of Neurology 2012; 72: 550-558)
2015 Identification of high prevalence of mild neurological variant of PMM2-CDG in Italy (J Neurol. 2015 Jan;262(1):154-64.)
2019 Participation International consensus guidelines for PMM2-CDG (J Inherit Metab Dis. 2019 Jan;42(1):5-28)
2020 Participation to the first clinical trial with Galactose in SLC35A2-CDG (Genet Med. 2020 Jun;22(6):1102-1107).
2021 Participation to International consensus guidelines for PGM1-CDG (J Inherit Metab Dis. 2021 Jan;44(1):148-163).
2021 Participation to Identification of the dominant variant of the STT3A gene (Am J Hum Genet. 2021 Nov 4;108(11):2130-2144.)

Funding and clinical trials (International)

2000-2005 EC: 5th framework 1999. Proposal No. QLRT-1999-00314; EC: A systematic approach towards the understanding, diagnosis and treatment of CDGS, a novel group of inborn metabolic disorders caused by defects of glycosylation». Role in the project: Investigator

2005-2008 EC:: 6th framework. EUROGLYCANET Coordination Action Proposal No.512131. Role in the project: Principal Investigator

2006 Multi Center, Multi National Open Label Extension Study for MPS VI ASB-03- 06 (local site-Dipartimento di Pediatria, Università di Catania). Role in the project: Investigator

2010-2015 MPS VI Clinical Surveillance Program (CSP) Observational Model: Cohort. Time Perspective. Role in the project: Principal Investigator

2013-2016 ERA-Net for Research Programmes on Rare Diseases: EURO-CDG A European research network for a systematic approach to CDG and related Diseases. Role in the project: Principal Investigator Italy.

2020-2023 Natural history study of PMM2-CDG. Role in the project: Principal Investigator Italy https://clinicaltrials.gov/ct2/show/NCT03173300.

Collaborations and Networks

European study group on Congenital Disorders of Glycosylations (Euroglycan, Euroglycanet, Euroglycan Omics) from 1999 to date.
Rare Diseases Clinical Research Network (RDCRN): Consortium "Frontiers in Congenital Disorders of Glycosylation" (US) from 2022.

Diagnostic Activity

Supervisor for the diagnosis of patients with neurodevelopmental disturbances (almost 500 novel patients per year), at the Child Neuropsychiatry Unit, University Hospital Catania.
Supervisor of the screening test (CDT) for the Congenital Disorders of Glycosylation (>700 samples per year) and consultant for the diagnosis of neurometabolic diseases at the referral center for inherited metabolic diseases, University Hospital Catania.

Membership

Member Society for the Study of Inborn Errors of Metabolism (SSIEM)
Member European Academy of Childhood Disability (EACD)
Member Italian Society for the study of inborn errors of metabolism (SIMMESN)
Member Italian Society of Child Neurology and Psychiatry (SINPIA)
Member of Scientific committee of the following Associations:
Congenital Disorders of Glycosylation Families and Professionals International Association
Italian CDG Association
Italian Association for Mucopolysaccharidosis and related disorders.
Italian Study Group of Joubert syndrome
Italian Study Group of Ceroidolipofuscinosis

Catania, 26 Apr 2022 Rita Barone