**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

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Scopus Author ID: 7006729370

ResearcherID: K-1412-2016

https://sciprofiles.com/profile/346245

**Present Position**

Associate 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

Adjunct 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, Aprile 2022): Number of publications 143, H-index 29; citations 2861.

**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:

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”.

2009Research 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

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 Scientific Achievements**

1996 Mapping of the locus of CDG-I (PMM2-CDG) (Genomics. 1996 Aug 1;35(3):597-9.)

1997 Identification of the phosphomannomutase deficiency as main cause of CDG-I (J Inherit Metab Dis. 1997 Jul;20(3):447-9)

1997 Characterization of the carbohydrate deficiency glycoprotein syndrome (J Med Genet. 1997 Jan;34(1):73-6. Syndrome of the Month)

1999 Participation to the CDG nomenclature assignation international workshop (Glyconj. J. 1999; 16: 669-671)

2012 Identification of the DPM2 gene (DPM2-CDG) (Annals of Neurology 2012; 72: 550-558)

2019 International consensus guidelines for PMM2-CDG (J Inherit Metab Dis. 2019 Jan;42(1):5-28)

2020 Clinical and biochemical improvement with galactose supplementation in SLC35A2-CDG (Genet Med. 2020 Jun;22(6):1102-1107).

2021 International consensus guidelines for PGM1-CDG (J Inherit Metab Dis. 2021 Jan;44(1):148-163).

2021 Identification of the dominant variant of the STT3A gene (Am J Hum Genet. 2021 Nov 4;108(11):2130-2144.)

**Funding (national)**



**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.

EURO-GLYCOTRAIN Marie Skłodowska-Curie Actions Doctoral Network (MSCA-DN) Horizon Europe 2023-2024.Role in the project: **“Investigator as: “Associate Partner University of Catania”. Submitted.**

**2019-2024** Natural History Study Protocol in PMM2-CDG (CDG-Ia) International, multicentric study

 ClinicalTrials.gov Identifier: NCT03173300

 https://www.clinicaltrials.gov/ct2/show/NCT03173300?cond=PMM2-CDG&draw=2&rank=3

Ruolo: **Principal Investigator**

**2022-2026** GM1 and GM2 Gangliosidosis PROspective Neurological Disease TrajectOry Study (PRONTO).

ClinicalTrials.gov Identifier: NCT05109793

<https://www.clinicaltrials.gov/ct2/show/NCT05109793?cond=Gangliosidosis&draw=2&rank=11>

 Ruolo**: Principal Investigator**

**2022-2024** Hunter Outcome Survey (HOS)

[ClinicalTrials.gov Identifier: NCT03292887](https://www.bing.com/ck/a?!&&p=d93ac336e8cdca5bJmltdHM9MTcwNDU4NTYwMCZpZ3VpZD0yZGU1OWIxMy01NmJjLTZkNDYtMmI2Ny04OGI5NTc2MTZjZWYmaW5zaWQ9NTIzMw&ptn=3&ver=2&hsh=3&fclid=2de59b13-56bc-6d46-2b67-88b957616cef&psq=Hunter+Outcome+Survey+(HOS)+clinical+trials&u=a1aHR0cHM6Ly9jbGFzc2ljLmNsaW5pY2FsdHJpYWxzLmdvdi9jdDIvc2hvdy9OQ1QwMzI5Mjg4Nw&ntb=1" \t "_blank)

<https://www.clinicaltrials.gov/ct2/show/NCT03292887?type=PReg&cond=Hunter+Syndrome&draw=2&rank=1>

Ruolo**: Principal Investigator**

**2022-2024** Age Group With Pompe Disease or With Mucopolysaccharidosis Type I (MPS I) in a Home-care Setting (HomERT)<https://www.clinicaltrials.gov/ct2/show/NCT05073783?cond=Pompe&cntry=IT&draw=2&rank=1>

**Ruolo: Principal Investigator**

**2023-2025** CDT-TCNPC-301 Studio di fase III, in doppio cieco, randomizzato, controllato- con placebo, a gruppi paralleli, multicentrico per valutare la sicurezza, tollerabilità ed efficacia di 2000 mg/kg di Trappsol® Cyclo™ (idrossipropil-β-ciclodestrina) e terapia standard rispetto a placebo e terapia standard in pazienti affetti da malattia di Niemann-Pick di tipo C1 – Codice Eudract:2020-003136-25 - Promotore :Cyclo Therapeutics

<https://www.clinicaltrials.gov/study/NCT04860960?intr=Trappsol%C2%AE%20Cyclo&rank=2>

**Ruolo: Principal Investigator**

**Altri trials clinici**

**2006-2009** Multi Center, Multi National Open Label Extension Study for MPS VI ASB-03- 06 Role in the project: **Investigator**

**2010-2015** MPS VI Clinical Surveillance Program (CSP) Observational

Time Perspective.

Ruolo : 2010-2014: **Investigator**.

2015: **Principal Investigator**

**2022-2024** A Multicenter, Multinational, Observational Morquio A Registry Study (MARS)

 https://www.clinicaltrials.gov/ct2/show/NCT02294877?cond=Morquio+Disease&cntry=IT&draw=2&rank=2Azafaros AZA-001-5A4-01 dal 01.11.2022

 **Ruolo: Principal Investigator**

**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.

**Professional Actvity**

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 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