

PERSONAL INFORMATION

Sabrina Rita Giglio

Full Professor of Medical Genetics – University of Cagliari
 Department of Medical Sciences and Public Health
 Director, Complex Structure of Medical Genetics, ASL8 Cagliari
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Sex Female | Nationality Italian

Current Position: FULL PROFESSOR, MED/03

Degree in Medicine and Surgery – University of Sassari (1993)

Specialisation in Medical Genetics – University of Pavia (1997)

ACADEMIC EDUCATION

PhD in "Human and Hereditary Pathology" – University of Pavia (2000)

Clinical internships in medical genetics, paediatric and adult genetic counselling, and cytogenetics (Universities of Sassari, Pavia, and Chicago)

 STUDY ABROAD EXPERIENCE

1998–2001: Department of Human Genetics, University of Chicago

 ACADEMIC POSITIONS

- Full Professor of Medical Genetics – University of Cagliari (since 2020)
- Director of the School of Specialisation in Medical Genetics – University of Cagliari
- Previously Associate Professor – University of Florence (2005–2020)
- Lecturer in undergraduate, postgraduate, and PhD courses at the Universities of Florence, Pavia, Milan, and Cagliari

 INTERNATIONAL TEACHING

2000–2001: Lecturer in Human Genetics for the course "Human Diseases and Phenotypes", PhD Programme in Human Genetics, University of Chicago

 CLINICAL AND HOSPITAL ACTIVITY

- From 16/02/2025: seconded to San Raffaele Hospital, Milan, as Director of Medical Genetics, Molecular Genetics and Cytogenetics
- 01/10/2020–present: Director, Complex Unit of Medical Genetics, Immunogenetics and Transplant Immunology, ASL8 Cagliari – P.O. Binaghi
- 2011–2020: Director, Complex Unit of Medical Genetics, Meyer Children's Hospital, Florence
- 2014–2015: Director, Complex Unit of Medical Genetics, Careggi Hospital, Florence
- 2004–2005: Medical Manager and Director of Clinical Genetics Laboratory, Meyer Hospital
- 2001–2004: Medical Geneticist and Head of Genetics Unit, San Raffaele Hospital, Milan
- 1997–1999: Clinical Genetic Counsellor at the University of Pavia and Pavia's ASL Genetic Counselling Centre

CLINICAL AND LABORATORY
ACTIVITIES

Prof. Giglio has over twenty years of specialised clinical experience in Medical Genetics, with particular expertise in genetic counselling pre- and post-test, both in prenatal and postnatal contexts for paediatric and adult patients.

She has developed multidisciplinary clinics and personalised diagnostic-therapeutic pathways for:

- Congenital malformations and complex genetic syndromes
- Skeletal dysplasias and collagenopathies
- Neurodevelopmental disorders
- Genetic nephropathies
- Inherited metabolic and endocrine disorders
- Rare paediatric and adult cancers
- Precocious puberty and monogenic obesity
- Hereditary eye diseases

She has directed Complex Units of Medical Genetics at national reference hospitals and restructured and enhanced clinical, diagnostic, and translational research services.

In laboratory management, she has overseen the shift from classical cytogenetic techniques to advanced NGS technologies, including:

- Array CGH/SNP
- FISH, PCR, methylation assays
- Clinical exome sequencing (50–60 cases/month)
- Whole-genome sequencing
- Liquid biopsy for solid tumours and inflammatory diseases
- cfDNA and dd-cfDNA analysis for transplant monitoring and prenatal screening
- Targeted panels for rare diseases such as genetic epilepsy, MODY, skeletal dysplasias, craniosynostosis, overgrowth syndromes, vascular anomalies, and complex malformative syndromes

Her laboratory is a national and international reference centre for molecular diagnosis of tubulopathies and genetic nephropathies.

She is highly experienced in genomic variant interpretation, ACMG/AMP-compliant classification, and integrated reporting for precision medicine.

<u>RESEARCH PROJECT LEADERSHIP</u>	
	2022–2025 PNRR – Heal Italia – Partnership in Diagnostics and Innovative Therapies in Precision Medicine, Spoke 1, Sardinian Population Sequencing (funding: €2,500,000)
	2022–2025 PNRR – M4 – C2 – INVESTMENT 1.5: eINS – Ecosystem of Innovation for Next Generation Sardinia, Coordinator of Spoke 10, Biopharmacology (funding: €5,000,000)
	2022–2025 Leader of the Operational Health Plan Project (POS), Trajectory 3: “Creation of a Precision Medicine Programme for the Mapping of the Human Genome on a National Scale” T3-AN-02 “NeurOmic.DataNet”: Genotyping to enhance predictive capabilities and diagnostic and therapeutic personalisation of two high-impact complex neurodegenerative diseases: Multiple Sclerosis and Parkinson’s Disease (funding: €7,000,000)
	2021–2024 Project Leader of the #DonAjò CAMPAIGN for the recruitment of bone marrow donors associated with the promotion of the regional bone marrow donor registry in Sardinia (funding: €150,000 per year)
	2021–2022 Project Leader – Fondazione Sardegna (ROL Call): Liquid biopsy for early diagnosis of liver cancers in Sardinia: how to prevent, save lives and reduce the economic burden on the healthcare system (funding: €75,000)
	2020 – Health Research Call 2018 (DD 15397/2018), NIKE Project (Managerial Decree No. 975 of 16/01/2020, published in BURT No. 06, Supplement 20, Part Three, dated 05/02/2020) (funding: €850,000). Research and Care for Patients Affected by Kidney Diseases
	2018–2020 Project RF-2016-02361560 “CHILDHOOD APRAXIA OF SPEECH: NEUROBIOLOGICAL AND BEHAVIOURAL MARKERS AND EXPERIENCE DEPENDENT CHANGES OF NEURAL CONNECTIVITY INDUCED BY TREATMENT”, in collaboration with Fondazione Stella Maris, Pisa
	2010–2011 AMLET – Androgens, Metabolism, Lifestyle, Environment: Testicular Function for Health (PRIN)
	2008–2011 Clinical and Genetic Study of Patients with Early-Onset Cryptogenic Epilepsy (PRIN)

<u>INSTITUTIONAL ACTIVITIES</u>	
	October 2024 – present: Member of the Technical-Scientific Committee (CTS) for Rare Diseases in Sardinia
	September 2023 – present: Scientific Consultant for the Italian Red Cross, Torrigiani Facility – Florence
	June 2023 – present: Representative of the Region of Sardinia for the census of genomic activities within the framework of the Inter-Institutional Coordination Table for Genomics in Public Health for the implementation of the National Plan for Health System Innovation
	2022 – present: Regional Coordinator of SIGU Sardinia
	2021 – present: Consultant for the spin-off PeopleCypher
	2019 – 2022: Member of the Scientific Committee of the Fiorenzo Fratini Foundation ONLUS
	2019 – present: Member of the Scientific Committee of the innovative start-up GENANDO Srl
	2012 – 2021: Member of the Scientific Committee of the Association for Childhood Kidney Diseases in Tuscany (A.Ma.R.T.I.)
	2015 – 2018: Regional Coordinator of SIGU Tuscany
	2013 – 2018: Member of the Scientific Committee of the FiorGen Foundation Onlus
	2008 – 2017: Programme Manager for “Laboratory Management: governance, activities and innovation” for the Tuscany Region, appointed

by Meyer Children's University Hospital within the framework of collaborations between the Meyer laboratory facilities and other Area Vasta structures, in accordance with regional guidelines

AWARDS AND RECOGNITION

- **2021:** Finalist in the final phase of the Start Cup Sardegna with the spin-off *PeopleCypher* and winner of first prize
- **2016:** Finalist of Start Cup Toscana with the project *Genando*
- **2002:** ESHG Strasbourg Award: *Heterozygous submicroscopic inversions involving olfactory receptor-gene clusters mediate the recurrent t(4;8)(p16;p23) translocation*
- **2001:** ICHG Vienna Award: *A submicroscopic chromosome inversion as the basis for two macroscopic chromosome rearrangement*
- **2000:** ASHG Philadelphia Award: *Olfactory receptor gene cluster mediate common chromosome rearrangement*

MEMBERSHIP IN SCIENTIFIC SOCIETIES

Member of the Italian Society of Human Genetics (SIGU), European Society of Human Genetics (ESHG), Italian Society of Obstetric and Gynaecological Ultrasound (SIEOG), American Society of Nephrology (ASN), European Cytogeneticists Association (ECA)

She has also participated as an invited speaker at numerous national and international conferences.

SUPERVISION OF THESES AND TUTORING

From 2001 to present, supervisor of several PhD theses and numerous experimental theses by students of the Degree Courses in Medical Biotechnology, Medicine and Surgery, Dentistry, Nursing Sciences, and numerous Specialisation theses in Medical Genetics and Nephrology.

She has also served as a committee member in various PhD programmes at the national level and in type A and B researcher recruitment competitions.

EDITORIAL BOARD – SCIENTIFIC JOURNALS

Member of the editorial board of:
Journal of Clinical Medicine, Obstetrics & Gynaecology Section
Review Editor for *Frontiers in Oncology*

SCIENTIFIC ACTIVITY

Author of around 200 publications in indexed international journals and of several book chapters in both English and Italian.

PERSONAL SKILLS

Communication Skills: Demonstrated strong verbal communication abilities through numerous presentations delivered to large audiences. Proven capacity to formulate and present proposals to senior management and to negotiate process improvements, both in academic and clinical settings.

Leadership Skills: Extensive experience in coordinating teams and leading projects, acquired during both university activities and within hospital environments.

Research Skills: Proven expertise and aptitude in conducting investigations, performing in-depth analyses, and exploring a wide range of processes with the objective of identifying and implementing effective improvements.

Organisational / managerial skills Presently coordinating a team of 40 staff members across laboratory, clinical, and administrative functions

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