

MD, full board in Medical Genetics, PhD in Human Genetic  
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Full Professor of Medical Genetics (MED/03) – School of Medical and Pharmaceutical Sciences, University of Genoa - Department of Neurosciences and Rehabilitation, Ophthalmology, Genetics, and Maternal-Child Sciences (DiNOGMI). Director of the Medical Genetics Unit of the IRCCS Policlinico San Martino in Genoa and of the Company Coordination Center for Rare Diseases.

Director of the Residency School in Medical Genetics.

Member of the Faculty of the PhD program in Neurosciences of DiNOGMI.

Member of the Council of the School of Medical and Pharmaceutical Sciences.

Member of the Board of DiNOGMI.

From 2013 to 2018 member of the Ligurian Regional Ethics Committee as an expert evaluator in medical genetics.

Member of the evaluation committee ASN 2019-2021 for the disciplinary sector MED/03.

From 2018 to 2025, a member of the Board of the Italian Association for the Study of Peripheral Nerve (ASNP).

From 2024, a member of the Board of the Italian Society of Human Genetics (SIGU).

### **Education and training**

- 1993 Specialization in Medical Genetics 50/50 with honors - University of Genoa - Genoa – IT
- 1991 PhD in Human Genetics - University of Turin - Turin – IT
- 1986 Degree in Medicine and Surgery 110/110 with honors - University of Genoa - Genoa – IT

### **Professional Training**

- Since December 2005: Full Professor of Medical Genetics (MED/03) at the Faculty of Medicine and Surgery of the University of Genoa
- Since April 2005: Director of the UOC Medical Genetics at the IRCCS Policlinico San Martino in Genoa
- Since November 2000: Associate Professor of Medical Genetics (MED/03) at the Faculty of Medicine and Surgery of the University of Genoa
- From August 1993 to October 2000: Senior Manager Level A at the Medical Genetics Service - Hospital Organization Ospedale S. Martino in Genoa and Affiliate University Clinics.

### **Educational Activities**

- Teaching of Medical Genetics (MED03) in the integrated course of Internal Medicine II for the Degree Course in Medicine and Surgery.
- Teaching of Medical Genetics at the School of Specialization in Medical Genetics.
- Teaching of Medical Genetics at the Schools of Specialization in: Pathological Anatomy, Child Neuropsychiatry, Nutrition Science, Diseases of the Digestive System, Diseases of the Cardiovascular System, Physical Medicine and Rehabilitation, Neurology, Dermatology and Venereology, Ophthalmology, Medical Oncology, Hematology.
- Lecturer of the Doctorate in Applied Neurosciences.
- Tutoring activities in the laboratory and in practice for PhD students in Applied Neurosciences and for residents in Medical Genetics.

### **Participation in scientific societies**

- since 1990 - Italian Association of Medical Genetics now Italian Society of Human Genetics (S.I.G.U)
- since 1996 – Italian Association for the Study of Peripheral Nerve
- since 2007 – Italian Society of Neurology (SIN)

### **Publications in internationally indexed journals (as of 30.05.2025)**

- HI= 38
- 211 publications in national and international scientific journals (Scopus)

The scientific activity has mainly focused on the study of neurodegenerative diseases (Parkinson's disease and parkinsonisms; Alzheimer's disease and other dementias; motor neuron diseases and other neuromuscular diseases in adults; cerebrovascular diseases). Neurodegenerative diseases, which represent one of the leading causes of morbidity and mortality in the elderly population, are the subject of research both through projects focused on each specific pathology and through broader projects aimed at studying the 'omics' of neurodegeneration (exome, genome, transcriptome, etc.) integrated with clinical characterization, imaging, and biomarkers of patients enrolled in the studies.

The main project is the NeurOmics project (Omics sciences for the identification of pathogenetic mechanisms and biomarkers in neurodegenerative diseases), which aims to use current knowledge and technological possibilities offered for the analysis of 'omics' sciences, from the sequencing of the human genome to the study of the proteins it encodes, in order to obtain a more complete characterization of patients based on the combination of omics/genomic and clinical data to radically improve the diagnosis, prognosis, and treatment of neurodegenerative diseases.

This project is conducted in collaboration with the center of the Italian Institute of Technology CMP3VdA and the IRCCS Cà Granda in Milan. Other research projects are conducted both within the IRCCS Neuroscience and Rehabilitation Network Projects (RIN) of the Virtual Institutes of Pathology (IV Movement Disorders, IV Dementias, IV Rare Neurological Diseases, IV Neuroimmunological Disorders, IV Motor Neuron Diseases, IV Epilepsies) as well as in projects of the PNRR. Additionally, clinical research includes participation in observational and interventional studies for innovative drugs for rare diseases.