Emilia Bellone - Curriculum vitae

Graduated in Biology, full board in Medical Genetics, PhD in Human Genetics
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Associate Professor in 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).
Member of the Faculty of the PhD program in Neurosciences of DINOGMI.
Member of the Teaching Commission of DINOGMI.
From 2013 to 2017 member of the Ligurian Regional Ethics Committee as an expert evaluator in medical genetics.
From 2013 to 2018 member of the Board of the Italian Association for the Study of Peripheral Nerve (ASNP).

Education and training

- 1996 Specialization in Medical Genetics 50/50 with honors - University of Genoa - Genoa – IT
- 1993 PhD Degree in Human Genetics - University of Turin - Turin – IT
- 1985 Master Degree in Biology 110/110 with honors - University of Genoa - Genoa – IT

Professional Training

- Since November 2017: Associate Professor in Medical Genetics (MED/03) at the Faculty of Medicine and Surgery of the University of Genoa
- Since November 2001: Assistant Professor of Medical Genetics (MED/03) at the Faculty of Medicine and Surgery of the University of Genoa
- Since November 2001: Biologist Manager at the Medical Genetics Service - Hospital Organization Ospedale S. Martino in Genoa and Affiliate University Clinics.

Educational Activities

- Professor of Medical Genetics (MED/03) at the Integrated Course of Medicine II for the Medical Degree Course, university of Genoa
- Professor of Medical Genetics at Nursing and Pediatric Nursing Degree Course, University of Genoa
- Professor of Medical Genetics at Degree Course in Health Professions, University of Genoa
- Professor of Medical Genetics at Specialization School in Medical Genetics, University of Genoa
- Professor of Medical Genetics at Specialization School in Child Neuropsychiatry, Ophthalmology, Hematology, Nephrology, Geriatrics, Psichiatry and Gynecology and Ostetrics.
- Lecturer of the Doctorate in Applied Neurosciences.
- Tutoring activities in the laboratory for PhD students in Neurosciences and for residents in Medical Genetics, University of Genoa.

Participation in scientific societies

- since 1994 - Italian Association of Medical Genetics now Italian Society of Human Genetics (S.I.G.U)
- since 1996 – Italian Association for the Study of Peripheral Nerv

Publications in internationally indexed journals (as of 30.05.2025)

- HI= 26
- 128 publications in national and international scientific journals (Scopus)

The scientific activity has mainly focused on the study of neuromuscular diseases of adult, genetically determined.
Neuromuscular diseases, which represent one of the leading causes of invalidity and mortality in the 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.
The research has in particular focused on Charcot-Marie-Tooh disease (CMT) and correlated disorders, including a broad spectrum of inherited diseases of Peripheral Nervous System (PNS) characterized by high clinical and genetic heterogeneity. Genetic test, after exclusion of the more frequent defect (CMT1A), relies on analysis by Next Generation Sequencing (NGS) of a CMT gene panel, reaching a genetic definition in 35% of the cases. In cases without diagnosis is possible a Whole Exome Sequencing (WES).
The research activity is focused on the identification of novel CMT-related genes and their pathogenetic mechanisms. This approach facilitates the identification of novel therapeutic strategies for these SNP disorders, which, given their prevalence in the population (1 in 2,500), represent a significant public health concern.
Indeed, the achievement of molecular diagnosis has been demonstrated to enhance the diagnostic pathway, thereby averting the prescription of inappropriate therapeutic interventions (e.g. orthopaedic surgery, immunosuppressive therapies) and enabling appropriate genetic counselling. This, in turn, allows patients to access prenatal/preimplantation testing and potential targeted therapeutic interventions (precision medicine).