BIOGRAPHICAL SKETCH

Marina Grandis

CURRENT POSITION
2021 –Associate Professor in Neurology; Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics and Maternal/ Child Sciences (DINOGMI); University of Genova, Italy.

EMPLOYMENT
2010-2021: Assistant Professor in Neurology (MED/26); Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics and Maternal/ Child Sciences (DINOGMI); University of Genova, Italy.
2007-2009: Research fellowships in the Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics and Maternal/ Child Sciences (DINOGMI); University of Genova, Italy.
2004-2006: Ph.D. program in Neurology, Psychiatry and Neurogenetics. University of Genova, Italy.
2002-2003: Research fellowship under the supervision of Prof. Michael Shy; Department of Molecular Medicine; Wayne State University; Detroit, MI, USA.
1997-2002: Postgraduate Residency in Neurology, University of Genova, Italy.

EDUCATION
2007 Ph.D in Neurology, Psychiatry, and Neurogenetics, University of Genova, Italy.
2002 Degree in Neurology (*cum laude*), University of Genova, Italy.
1997 Italian Board of Medical Doctors, University of Genova, Italy.
1996 Medical Doctor Degree (*cum laude*), University of Genova, Italy

RESEARCH EXPERTIES
Cell biology: establishment of primary Schwann cell cultures from MPZ knock out mouse; transfection of cell lines and primary Schwann cell cultures; intercellular adhesion assays.
Animal models: manipulations of laboratory mice (genotyping, electrophysiology, behavioural tests).
Molecular biology: In vitro-mutagenesis. DNA, RNA and Protein extraction from different sources (tissues and cells) and processing (Semiquantitative RT-PCR, Real-Time PCR, Western Blot).
Light microscopy: tissues and cultures processing for morphological and morphometrical evaluations (immunohystochemistry, immunofluorescence, image analysis using the Image Pro-Plus Software).

CLINICAL EXPERTIES
2004- Consultant Neurologist at Peripheral Neuropathies Outpatient Clinic.
2004- Consultant Neurologist at CMT Outpatient
2011- Consultant Neurologist at the IRCCS AOU San Martino-IST in Genova
2013- Consultant Neurologist at Neuromuscular Outpatient Clinic
2016- Consultant Neurologist at TTR-FAP Outpatient Clinic

PERSONAL STATEMENT
I am a Neurologist with a long experience in neuromuscular diseases including inherited neuropathies, inherited and inflammatory myopathies and Myasthenia Gravis.
My research activity and clinical interests are mainly focused on the pathogenetic mechanisms, diagnosis, and therapy of neuromuscular diseases.

Myopathies
During the last ten years I started an outpatient clinic dedicated to adult muscular diseases, with a main focus on genetic forms.
Since 2014 I am a member of the Italian Association of Myology, and I have been included in several Italian collaborative projects mainly focused on genetic myopathies. Since 2025 I am part of the board of the Italian Society of Myology.

Moreover, I developed a diagnostic algorithm for the diagnosis of adult patients with persistent hyperckemia, using electromyography, muscle MRI and genetic screening, including MLPA and NGS approaches. This diagnostic algorithm has been extended to other Italian centres (Prof. Tiziana Mongini and Dott.ssa Lucia Ruggiero), proving to be useful to reach a diagnosis in a consistent percentage of patients. The results have been published in 2020 (Gemelli et al; Muscle & Nerve).
I participated also to collaborative projects on Pompe disease and, since 2022, I participate to the Italian Clinical Group FSHD meetings, sharing MRI, genetic and clinical data with the main Italian centres.
In particular, I am the PI for the Genoa center of a Telethon research grant dedicated to FSHD: "Natural history and biomarkers in Facioscapolo-Humeral Muscular Dystrophy", coordinated by Prof. Massiiliano Filosto, which has been funded in 2024.

Inherited Neuropathies
In 2002 and 2003, I served as a Research Associate at the Department of Molecular Medicine at Wayne State University (Detroit, MI, USA), in the laboratory of Prof. Michael Shy. During this year I was involved in CMT1B and the different pathomechanisms underlining the diverse clinical phenotypes.
During my Ph.D. in Neurosciences (2004-2006), I studied the pathomechanisms underlining the two main phenotypes of P0-related neuropathies. In particular, I demonstrated that *MPZ* mutations causing an early onset phenotype may be alternatively mistraffricked with a frequent entrapment in the ER or correctly expressed into the plasma membrane where they disrupt the intercellular adhesion function (Human Molecular Genetics 2008).
Since 2004, I contributed to establish, along with Prof Angelo Schenone, Prof. Paola Mandich and Prof. Emilia Bellone the Integrated Clinic for Hereditary Neuropathies in Genoa. Several publications have emerged from the clinical activities of this clinic, and the Genoa Center was selected to participate in major international trials for CMT1A, including the "multicenter, randomized, double-blind, placebo-controlled trial of long-term ascorbic acid treatment in Charcot-Marie-Tooth disease type 1A (CMT-TRIAAL)" (results published in Lancet Neurology) and the Pharnext trial "CLN-PXT3003-06 where I served as PI for the Genoa Center.
Since 2008, I focused my interest on the role of altered glycosylation as a pathogenetic mechanism in neuropathies caused by MPZ mutations. As part of this project, I collaborated with Dr. Maurizio D'Antonio (San Raffaele Hospital, Milan) and Prof. Jean Michel Vallat (Centre Hospitalier Universitaire de Limoges) for the development and phenotypic characterization of a murine animal model of MPZ mutation (P0D61N) corresponding to a hyperglycosylated form of P0. This collaboration led to an application for a grant from AFM-Téléthon, which was funded and allowed the creation of the first knock-in animal model expressing a P0 mutation along with the wild-type allele, accurately recreating the human genotype without the deleterious effects of P0 overexpression. The results have been published in Human Molecular Genetics Journal in 2022.
Currently, in my lab, we are working on RNA interference to silence the mutated allele carrying the pathogenic MPZ variant, in order to rescue the neuropathic phenotype.

Myasthenia Gravis
I am also interested, as a clinician, in Myasthenia Gravis (MG) and I have been involved as local PI in several clinical trials.
Since the last years I am part of the Italian Study Group on Myasthenia Gravis and I collaborate to the project of an Italian Registry on patients affected by Myasthenia Gravis.
Further, my group has been involved in clinical studies concerning MG related to cancer immunotherapy and we are currently working on a systematic review on Myocarditis-myositis-myasthenia complex in patients treated with immunotherapy.

GRANTS

- 2024- Partner in Telethon GUP24010M: Natural history and biomarkers in Facioscapolo-Humeral Muscular Dystrophy.
- 2022- A Multiscale integrated approach to the study of the nervous system in health and disease (MNESYS).Role: leader of WP4 (spoke 6). Funding Source: #NextGenerationEU (NGEU) funded by the Ministry of University and Research (MUR), National Recovery and Resilience Plan (NRRP).
- 2022-2024: The complexity of diagnosis and the innovative therapies for neuromuscular diseases. Italian Ministry of Health – Ricerca Corrente.
- 2018-2021: Inherited neuromuscular diseases, from clinical evaluation to novel diagnostic, radiological and molecular approaches. Italian Ministry of Health – Ricerca Corrente.
- 2017-2018 Principal Investigator in AFM-Telethon (20572): Misglycosylation in Charcot-Marie-Tooth neuropathies associated to MPZ mutations.
- 2016-2017. Partner in Telethon GUP15010: TTR-FAP Italian Registry: a collaborative network for definition of natural history, psychosocial burden, standards of care and clinical trials.
- 2012-2013. The role of Misglycosylation in CMT neuropathies with particular focus on Myelin Protein Zero related neuropathies. University of Genova.
- 2010-2011. Study of Myelin Protein Zero (MPZ) mutations introducing new N-glycosites: pathomechanisms and therapeutic implications. University of Genova.2017-2018 Principal Investigator in AFM-Telethon (20572): Misglycosylation in Charcot-Marie-Tooth neuropathies associated to MPZ mutations.

HONORS AND AWARDS

Since May 2024 I am part of the Peripheral Nerve Society Scientific Program Committee, chaired by Kathrin Doppler and Vincent Timmerman.

2022-Member of the new management group del neuropathy working group- Euro-NMD ERN, coordinated by Dr. Davide Pareyson.

2021-Member of Board of Directors the National Virtual Rare Diseases Institute - The Italian Neuroscience and Rehabilitation Network (RIN).

2001 Best contribution at the Meeting of the Italian Peripheral Nerve Study Group (Sondrio, Italy).

2007 Best contribution (ex-aequo with 2 other participants) to the meeting of the Peripheral Nerve Society (Snowbird, Utah, USA).

CURRENT MEMBERSHIPS IN SCIENTIFIC SOCIETIES:
Italian Society of Neurology (SIN);
Italian Society Peripheral Neuropathies (AINP);
Italian Society of Myology (AIM);
International Peripheral Nerve Society (PNS).

BIBLIOMETRY
133 papers in international peer-reviewed journals.
H-index: 30.
Number of citations: 3721 (*Scopus, 9 June 2025*).

Genoa, December 9th, 2025