

## PERSONAL INFORMATION

### Paola Saveri

📍 IRCCS Foundation Carlo Besta Neurological Institute  
Rare Neurodegenerative and Neurometabolic Diseases Unit  
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## WORK EXPERIENCE

31<sup>st</sup> December 2019 to present

### Healthcare Biologist

IRCCS Foundation Carlo Besta Neurological Institute, Milan

Rare Neurodegenerative and Neurometabolic Diseases Unit

Research in the field of Charcot-Marie-Tooth disease (CMT) and related peripheral neuropathies with a particular focus on axo-glial interactions and CMT pathomechanisms. Preparing and conducting clinical experiments in hereditary neuropathy. Analysing and updating CMT patients' databases.

May 2014-December 2019

### Biologist (Co.co.co.)

IRCCS Foundation Carlo Besta Neurological Institute, Milan

Rare Neurodegenerative and Neurometabolic Diseases Unit

Characterization of patients affected by CMT or familial amyloidotic polyneuropathy from both a genetic (mutational studies) and biological viewpoint (myelin protein analysis and misfolded proteins' degradative pathways of nerve tissue). Analysing and updating CMT patients' databases.

March 2014-August 2014

### Internships

University College London Hospitals, London (UK)

Institute of Neurology, Neurogenetics Unit

Next generation sequencing for the identification of mutations related to peripheral neuropathies.

October 2012-April 2014

### Biologist (Fellowship)

IRCCS Foundation Carlo Besta Neurological Institute, Milan

Rare Neurological Diseases of Adulthood Unit

Support activities for genetic research in inherited neuropathies. Study of clinical case histories of patients affected by hereditary illness and rare dysmetabolic syndromes.

September 2011-April 2012

### Research technician

University at Buffalo, Buffalo (USA)

Jacobs School of Medicine and Biomedical Sciences, Hunter James Kelly Research Institute

Biology and pathophysiology in peripheral neuropathies in Krabbe disease.

February 2007- August 2011

### Research technician

San Raffaele Center Foundation, Milan

Biology of Myelin Unit

Axo-glial interactions in nerve development and pathology: the study of murine transgenic models of hereditary neuropathies.

## EDUCATION AND TRAINING

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- October 2005 **Master's in Biotechnological Methodology for the Environment**  
University of Perugia
- 1<sup>st</sup> session 2004 **Professional Biologist Qualification**  
University of Perugia
- July 2003 **Master's Degree in Biological Sciences**  
University of Perugia

## ACHIEVEMENTS AND AWARD

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- Grants** Co-investigator "Unravelling mechanisms of axonal loss in late-onset genetic neuropathies", Grant AFM Téléthon 20667
- Co-investigator "Mechanisms of axonal degeneration in late onset CMT1B neuropathies: molecular pathways and therapeutic approaches", Grant Telethon GPP 19099
- Co-investigator "ACT-CMT, Accelerate Clinical Trials in Charcot-Marie-Tooth Disease ", Grant NIH 8301
- Co-investigator "3-Genes, CMT1B USA-CMTAss, extension for CMT2A and CMTX1"  
University of Iowa Health Care

## TECHNICAL SKILLS

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Bacterial cultures, transformation, DNA purification; genomic and cDNA cloning and analysis; Southern Blot; RNA purification from cells and tissues; RNA reverse transcription; *in vitro* site-directed mutagenesis; PCR; primer design; Sanger DNA sequencing; quantitative PCR (Taqman); Next generation sequencing (targeted sequencing). Protein purification from cells and tissues; immunoprecipitation; Western Blot; immunocytochemistry and immunohistochemistry. Nerve teasing; use of cryostat; confocal microscopy. Cell cultures (BV-2, COS-7, MDCK, SH-SY5Y), transient transfection of eukaryotic cells; murine embryonic stem cells cultures. Mouse colony management and breeding strategies, genotyping, tissues dissection, motor tests (Rotarod, Grid test). Use leading software for acquisition and analysis imaging, bioinformatic instruments for the study of nucleic and amino acid sequences and for the analysis *in silico* of the sequence variants.