PERSONAL INFORMATION

Raffaella Brugnoni

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WORK EXPERIENCE 31/12/2019 - 30/12/2024 Italian NHS at the FINCB. Analysis by Next Generation Sequencing of two panels of genes associated with neuromuscular channelopathies and of the neuromuscular junction disease. Project in the field of pharmacogenomics, for the search for SNPs that can lead to a different response to drug treatment with mexiletine in patients suffering from non-dystrophic myotonias or myotonic dystrophies. 15/03/2019 - 30/12/2019 Grant RF189, FINCB. Project title: "Identification of B cell-related biomarkers to predict response to immunosuppressive and B cell targeting therapies in Myasthenia Gravis". 04/12/2017 - 03/03/2019 Grant TRANS-ALS-RA90, FINCB. Project title: "Translating molecular mechanism into ALS risk and patient's well-being - TRANS ALS." 29/12/2016 - 28/09/2017 Grant RF164-RF181, FINCB. Project title: "Assessing the immunomodulatory effect of probiotics in the Experimental Model of Myasthenia Gravis". - "High-throughput miRNAome profiling in myasthenia gravis patients: towards the identification of pathogenic mechanisms underlying autoimmunity". Grant FINCB. Project title: "Genetic analysis of pathological polymorphisms in muscle ion channels 16/05/2015 - 15/05/2016 responsible for genetic myopathies". Grant FINCB. Project title: Implementation of molecular diagnostics on channelopathies, congenital 11/03/2014 - 10/03/2015 myasthenic syndromes and congenital myopathies 16/02/2013 - 15/02/2014 Grant FINCB. Project title: "Implementation of the Molecular Diagnostics of Muscular Canalopathies and Myopathies". 01/02/2010 - 31/01/2013 Grant FINCB. Project title: "Scientific diagnostic activity at the Unit Neurology 4". 01/02/2005 - 31/01/2010 Permanent contract. Società consortile "NBS Biotech", c/o FINCB. Head of the Company's • laboratory. 1999 - 2005 3 grant of collaboration, FINCB, Project title: 1) Messa a punto di screening diagnostico rapido per mutazioni puntiformi in malattie neurologiche degenerative e organizzazione di DB Clinico-biologico. 2)Malattie autoimmuni del SNC e SNP: aspetti immunopatologici. 3) Studio delle molecole effettrici e regolatorie responsabili della perdita di tolleranza immunitaria nella Miastenia Grave e della reazione citotossica nella encefalite di Rasmussen e miopatie infiammatorie. 1996 - 1999 3 fellowship Senior FINCB, Project title: 1) Meccanismi effettori e regolatori nella patogenesi delle malattie autoimmuni del sistema nervoso centrale e periferico. 2) Patogenesi immuno-mediata nelle malattie autoimmuni e degenerative del sistema nervoso centrale e neuromuscolare. 3) Caratterizzazione clinica e correlazione fenotipogenotipo nelle malattie neuromuscolari ereditarie. 2 fellowship Junior FINCB, Project title: 1) Immunoterapia della miastenia grave: studio 1994 - 1996clinico biologico. 2) Myasthenia Gravis and EAMG: molecular characterisation of TCR and MHC-class II restriction of AChR-specific T lymphocytes. 1993 - 1994 **Training FINCB EDUCATION AND TRAINING** 1995 until now Registration in the professional register of the National Order of Biologists 1994 State examination for the qualification to the profession of Biologist, University of Milan 1993 Degree in Biological Sciences at the University of Milan, Thesis title: Preparation of molecular species of Sphingomyelin labelled with tritium at position 3 of Sphingosine.

ACHIEVEMENTS AND AWARD editorial activity	Reviewer of scientific articles: DMPI, Frontiers, Neuromuscular disorders, Pediatrics, Genes.
TEACHING ACTIVITY 2021-2022	Tutor for a master's degree at the Faculty of Biotechnology of the University of Verona and Biology at the Bicocca University of Milan.
TECHNICAL SKILLS	Development of molecular-diagnostic screening using next-generation sequencing (NGS) panels, for the search for pathological mutations and new disease genes. Knowledge of the following molecular biology methods: NGS and Sanger sequencing. Analysis of fragments using the latest generation of capillary sequencers. Genomic amplification by PCR and multiplex PCR. Knowledge of MLPA, Southern blot, RFLP, and SSCP techniques. Genomic DNA extraction from PBL, muscle and chorionic villi. RNA extraction, RT-PCR, FICOL and lymphocyte immortalization. Excellent knowledge of the following instrumentation: MiSeq, capillary sequencer mod. ABI3500dx, ABI3100 and ABI3130xl, ABI3900 oligonucleotide synthesizer, HPLC and DHPLC.