Trieste, 12 gennaio 2026

CURRICULUM VITAE of Massimo Zeviani

DOB 05/11/1955 in Genova, Italy

Resident in Padova via Schumann 5, 35132

Married, three children

Career/Employment

2022-present: Scientific Director of the Institute for Maternal and Child Health IRCCS Burlo Garofolo, Trieste Italy
2019-2022: Professor of Neurology, University of Padova
2013-2019: Professor of Mitochondrial Medicine, University of Cambridge UK, and Director of the MRC Mitochondrial Biology Unit, University of Cambridge, UK.
2011-2012: Director of the Department of Molecular Medicine, Istituto Neurologico "Carlo Besta", Milan
2003-2012: Director of the Unit of Molecular Neurogenetics at the Istituto Neurologico "Carlo Besta", Milan
1998-2002: Director of the Unit of Biochemistry and Genetics at the Istituto Neurologico "Carlo Besta", Milan
1996-1997: Director of the Unit of Molecular Medicine at the Children's Hospital "Bambino Gesù" in Rome (Italy)
1993-1996: Associate of Neurology at the Istituto Neurologico "C. Besta", Milan
1990-1993: Director of the Laboratory of Molecular Pathology of the Istituto Neurologico "C. Besta", Milan

Research interests and career highlights

Dr. Zeviani has identified and characterized numerous disease genes associated with OXPHOS defects helping to elucidate the molecular pathogenesis of mitochondrial diseases. To give a few examples: In 1993−1994 he spent a sabbatical year at INSERM Unit 393 directed by Prof. A. Munnich at the Hopital Necker−Enfants Malades in Paris, where he collaborated with Dr. Judith Melki in the identification of SMN, the gene responsible for spinal muscular atrophy (SMA). In collaboration with Andrea Ballabio (TIGEM) he found paraplegin, a component of the mitochondrial protein quality control pathway, as responsible of SPG7, a recessive form of hereditary spastic paraplegia. In 1998 he discovered Surf1 as the first assembly factor to be mutated in Leigh's syndrome associated with COX deficiency. In collaboration with A. Suomalainen, Helsinki University, Finland, he discovered the first mutant gene associated with autosomal dominant PEO, ANT1. The first mutation in the 18kDa subunit of complex I was also found and characterized in an infant patient in collaboration with Hans Spelbrink and Howy Jacobs, Tampere University, Finland. The discovery of pol−gammaA as a major disease gene in mitochondrial disorders, prompted us to proceed to a systematic investigation on the phenotypic and biochemical features of the conditions associated with pol−gamma A mutations, which produced a series of papers illustrating the spectrum of clinical and biochemical presentations associated with mutations in this gene. Additional results of this work have been the discovery of PUS1, EFTu and EFG1 mutation in mtDNA translation defects, ETHE1 as the gene responsible of ethylmalonic encephalopathy, of Mpv17 as responsible of a hepatocerebral mtDNA depletion syndrome, the molecular characterization of MR−1 in non−kinesigenic dyskinesia, FASTKD2 as responsible of an encephalopathy with COX deficiency, SDHAF1 as the first assembly factor of complex II, again responsible of an infantile leukoencephalopathy, AIF, apoptosis inducing factor, as responsible of an X−linked mitochondrial infantile encephalomyopathy1. The effort to create in vivo models of OXPHOS disorders has led us to obtain a Surf1 KO mouse model, a Mpv17 KO mouse model a SCO2KO/KI mouse model (in collaboration with EA Schon), and several additional models in mice and flies. An ETHE1 KO recombinant mouse allowed us to dissect the molecular defect of ethylmalonic

Encephalopathy and understand the role of the mutant gene product. Strong collaborations with Ileana Ferrero and her group on yeast modeling of human mitochondrial disorders, and with Rodolfo Costa for fly models, have both led us to widen the spectrum of in vivo experimental investigation on the pathogenesis of a number of OXPHOS defects due to nuclear gene mutations. More recently, he identified other disease-genes (APOPT1, PITRM1, TTC19, APOO, TIMM50 RNase H1 and many others). More recently, his lab's efforts have focused on developing therapeutic approaches to treat these conditions in experimental models and in patients. He explored several approaches to the therapy for mitochondrial diseases. These include the increase of mitochondrial biogenesis by stimulating the PGC1alpha pathway, AAV-based gene therapy to re-express the missing/mutated gene, the use of rapamycin to clear dysfunctional mitochondrial via autophagic pathway in mice, the bypass of respiratory chain block by using the alternative oxidase.
In addition to research activities, we are also committed to offer a complex and wide panel of biochemical and molecular assays for the diagnosis of mitochondrial and movement disorders, as well as a clinical care for outpatients and inpatients with suspected metabolic and mitochondrial dysfunction.

INDEXES
Dr. Zeviani is Author of 532 *in extenso* publications (articles, reviews, letters, book chapters).
Dr. Zeviani H-index is 129.

FELLOWSHIPS AND AWARDS
2013 “Prix de la Fondation NRJ 2013” (Institut de France) «Génétique des maladies dégénératives», France
2009 Gaetano Conte” Prize of the Mediterranean Society of Myology (Nicosia, Cyprus).
2008 The Sir William Dunn Scholars' programme MBU, MRC Cambridge, UK
2004 “René Descartes” EU award for European transnational research, EU

Ongoing Research Support

Title: Experimental gene therapy in mitochondrial disorders. (300K€)
Duration: 2021-2023
Granting Agency: Telethon (Project 19007)
Role: PI

Title: Experimental strategies to combat Pearson’s syndrome (210K€)
Duration: 2020-present
Granting Agency: Fondazione Luigi Comini Onlus
Role: PI

Title: Unravelling the function of Mpv17 (160K€)
Duration: 2023-2025
Granting Agency: Telethon (Project GMR23T1065)
Role: PI
EMBO Mol Med. 2019 May;11(5):e9561

Yours faithfully
Massimo Zeviani