

FABRIZI GIAN MARIA

Born on June 22 1959, Siena (Italy)

CURRENT POSITION

Associate Professor of Neurology at the Department of Neurosciences, Biomedicine and Movement, University of Verona (Italy). Neurologist, Chief of the Clinical Neurobiology Unit at the Department of Neurosciences, University Hospital G.B. Rossi, AOUI of Verona

EDUCATION

1984. Medical School degree (summa cum laude), University of Siena. Degree thesis: Schwann cell cultures from the Trembler mouse, a model of Dejerine-Sottas disease

1987-1989. Post-Doctoral Research Fellowship, H. Houston Merritt Clinical Research Center for Muscular Dystrophy and Related Diseases (Director Prof. S. Di Mauro), Columbia University College of Physician and Surgeons, New York, New York USA. Research activities: molecular biology of mitochondrial disorders

1990. Ph.D. ("Dottorato di Ricerca") in Anthropology, Anthropometry and Physiology of the Sport, University of Siena

1990. Specialization in Neurology (summa cum laude), University of Siena

PROFESSIONAL APPOINTMENTS AND ACADEMIC CAREER

1985-1986. Medical Officer in the Italian Army

1990-1996. Assistant Professor of Neurology and Neurologist, Department of Neurological Sciences (Chairman Prof. G.C. Guazzi), University of Siena

1996-2002. Assistant Professor of Neurology and Neurologist, Department of Neurological and Visual Sciences, (Chairman Prof. N. Rizzuto), University of Verona.

From 2002: Associate Professor of Neurology and Neurologist, University and AOUI of Verona.

RESEARCH INTERESTS AND SCIENTIFIC CONTRIBUTIONS

Molecular genetic and pathology of inherited and degenerative neurological disorders: inherited neuropathies; mitochondrial encephalomyopathies; degenerative dementias; motor neuron disorders. Trials aimed at developing clinical and instrumental outcome measures and therapies for inherited neuropathies.

Major scientific contributions:

- Cloning and characterization of nuclearly encoded subunits (subunits VIa, VIIa, VIIc, VIII) of human cytochrome c oxidase.
- Identification of the molecular basis of PMP2—related Charcot-Marie-Tooth disease type 1A and Hereditary Neuropathy with Liability to Pressure Palsies: definition of gain-of-function versus loss-of-function mechanism at the transcriptional level.
- Definition of the pathological basis of Charcot-Marie-Tooth disease type 2E: identification of giant-axons as the pathological hallmark of NEFL-related CMT.
- Contribution to the full characterization of phenotype-to-genotype spectrum of maternally-inherited mitochondrial disorders, Charcot-Marie-Tooth disease and related neuropathies, familial Alzheimer disease and Fronto-Temporal Dementias.

SCIENTIFIC SOCIETIES

2016: President-elect of the Italian Peripheral Nerve Society

Affiliations: Italian Neurological Society (SIN). Italian Peripheral Nerve Society (ASNP). Italian Society of Neuropathology (AINP) and Clinical Neurobiology (CN). International Consortium of Charcot-Marie-Tooth. Peripheral Nerve Society.

GRANTS AND RESEARCH PROJECTS

- 1992-1994: Collaborator, Telethon Project "Molecular characterization of new mitochondrial encephalomyopathies" (Principal Investigator Prof. G.C. Guazzi)
- 1993-1995: Principal investigator, Telethon Project "Clinical, neuropathological and molecular-genetic characterization of autosomal dominant ocular myopathies"
- 1996-1999: Collaborator, Telethon Project "Towards an etiological and pathogenetic diagnosis of the inherited neuropathies. A genotype and phenotype investigation of 200 patients with pathologically characterized nerve biopsies". (Principal Investigator Prof. N. Rizzuto)
- 1997-1999: Principal investigator, Project funded by Regione Veneto (Italy): "Clinical and Molecular-Genetic Study of the Fragile-X"
- 1998-2000: Collaborator, PRIN project #9806409869 by MIUR "Hereditary Neuropathies: physiopathology and molecular bases". (Coordinator Prof. N. Rizzuto)
- 2000-2002: Collaborator, PRIN project #MM06402489 by MIUR "Hereditary Neuropathies: identification of new genes, molecular and cellular pathology" (Coordinator Prof. N. Rizzuto)
- 2001-2003: Principal investigator of Research Unit, Project by the Italian Ministry of Health "Molecular analysis and pathogenetical models of Hereditary Motor-Sensory Neuropathies in Italy" (Coordinator Dr. F. Taroni)
- 2002-2004: Collaborator, PRIN project #2002061843 by MIUR "Hereditary Neuropathies caused by molecular lesions of myelin proteins: gene dissection, molecular pathology and pathogenetical mechanisms of secondary axonal degeneration" (Coordinator Prof. N. Rizzuto)
- 2003-2005: Collaborator, Telethon-UILDM project #GUP02169 "Quality of life and disability in patients with Charcot-Marie-Tooth disease. A Multicentre and multiperspective follow up" (Coordinator Dr. L. Padua)
- 2005-2008: Principal investigator of Research Unit, Telethon-UILDM project #GUP04002 "Multicentre, randomised, double-blind, placebo controlled trial of ascorbic acid treatment in Charcot-Marie-Tooth disease type 1A (CMT1A). (Coordinator Dr. D. Pareyson)
- 2005-2007: Principal investigator Research Unit, Telethon-UILDM project "Severe Charcot-Marie-Tooth disease and related hereditary neuropathies: an Italian collaborative network for implementing the molecular analysis of rare forms and new genes" (Coordinator Dr. F. Taroni).
- 2005-2007: Collaborator, Fondazione Mariani project #R-05-44 "Severe childhood-onset Charcot-Marie-Tooth disease: a collaborative project for implementing the molecular analysis of new genes" (Coordinator Prof. N. Rizzuto)
- 2005-2007: Collaborator, PRIN project #2005060584 by MIUR "Charcot-Marie-Tooth disease: characterization of novel genes and strategies of molecular therapeutics". (Coordinator Prof. N. Rizzuto)
- 2010-12 Principal investigator, Telethon-UILDM project GUP09013. "A Multicenter study to evaluate the effects on Charcot-Marie-Tooth neuropathy type 1A of a composite Treadmill, Stretching and Proprioceptive Exercise (TreSPE) rehabilitation program" (Coordinator Prof. A. Schenone)
- 2010-13 Principal investigator of Research Unit, Project by Cariverona "Cognitive and behavioural disability in dementias and psychoses: diagnostic and prognostic biomarkers of Mild Cognitive Impairment (Coordinator Prof. N. Rizzuto)
- 2011-13 Principal investigator, Telethon-UILDM project GUP 10008 "Novel outcome measures for Charcot-Marie-Tooth disease" (Coordinator Prof. G. Vita)
- 2013-15 Principal investigator of Research Unit, Telethon-UILDM project GUP 13006 "CMT National Registry: towards definition of standards of care and clinical trials" (Coordinator Dott. D. Pareyson)
- 2016 Principal investigator of the project "Implementation of a Biobank for inherited neuropathies, dementias and motor-neuron disorders" financed by the AOUI Verona.

2014-2016 Principal Investigator of Research Unit, Multi center study “Tafamidis – Neurological data collection protocol” (Coordinator Dott. D. Pareyson)

2016-2018 Principal investigator of Research Unit, project Telethon_UILDMD GUP15010 “TTR-FAP Italian Registry: a collaborative network for definition of natural history, psychosocial burden, standards of care and clinical trials” (Coordinator Prof. G.Vita)

2016-2018 Principal Investigator of research unit, multicenter project financed by Pfizer “Carpal tunnel syndrome: are there neurophysiological and ultrasound markers suggestive (or predictive) of Transthyretin amyloidosis? A cross-sectional and longitudinal multicenter study” (Coordinator Prof. Briani C.).

REVISION AND EDITORIAL DUTIES

Ad hoc reviewer for the following journals: Acta Neuropathologica, Annals of Neurology, Brain, Clinical Neurology Neurosurgery, European Journal of Neurology, Journal of Neurology, Journal of Neurological Sciences, Journal of Neuroscience Methods, Journal of Peripheral Nervous System, Muscle Nerve, Neurological Sciences, Neuromuscular Disorders.

2015: Associate Editor, Journal of Alzheimer’s disease.

PUBLICATION AND BIBLIOMETRICS

Prof. Fabrizi authored more than 120 publications on journals with international relevance and impact factor..

Scopus: H-index = 28

Web-of-Science: H-index = 50; average citation per item 26.21; citing articles =8741; citing articles without self-citations = 8541

Google-Scholar: H-index= 30; I-index=61; 3654 citations

Il/La Sottoscritto/a, ai sensi degli artt. 46 e 47 D.P.R. n. 445/2000, consapevole delle sanzioni penali previste dall’art. 76 D.P.R. n. 445/2000 nel caso di mendaci dichiarazioni, falsità negli atti, uso o esibizione di atti falsi o contenenti dati non più corrispondenti a verità, dichiara che quanto sopra riportato corrisponde a verità. Dichiaro inoltre che i titoli e gli allegati sono, su richiesta, disponibili in copia fotostatica conforme agli originali.

Autorizzo il trattamento dei miei dati personali ai sensi del Dlgs 196 del 30 giugno 2003”

Verona, February 19 2017

