CV Valeria Tiranti, PhD

Education

University of Milan − Biology degree cum laude: 1989.
University of Milan − Specialization in Medical Genetics cum laude: 2001.

Present Position

Permanent position as Dirigente Biologo, Unit of Molecular Neurogenetics, Foundation IRCCS Neurological Institute C. Besta

Research Interests

Dr. Valeria Tiranti has a long−standing expertise in mitochondrial and metabolic disorders, including identification of disease genes, biochemical evaluation of the respiratory chain activities, molecular and cellular biology, and identification of pathogenetic mechanisms of diseases. In addition, she recently joined the field of Neurodegeneration with Brain Iron Accumulation (NBIA) by characterizing the C19orf12 protein and by performing a metabolomics study on a cohort of patients with Pantothenate Associated Neurodegeneration (PKAN syndrome). She has been studying Pank2−KO mice, as well as patient's derived fibroblasts available at the Unit of Molecular Neurogenetics of Foundation IRCCS C. Besta. She is interested in understanding the role of mitochondria and iron in neurodegeneration by studying cellular and animal models in order to develop potential therepuctic intervention.

Organization

Foundation Neurological Institute C. Besta is an internationally recognized leading Centre in neuroscience; it diagnoses and treats neurological diseases in adults and children and carries out basic and clinical research in Neurology. It is composed of a Department of Clinical Neurology (250 beds), two Units of Child Neurology (40 beds), a Department of Neurosurgery (90 beds) and two Units of Neuroradiology and Radiotherapy. In addition a Department of Experimental Neurosciences includes the Unit of Molecular Neurogenetics, the Unit of Biochemistry and Genetics, the Unit of Neuromuscular Diseases, the Unit of Experimental Neurophysiology, and the Unit of Neuropathology. More than 25000 patients/year are examined and treated at the Institute. The Unit of Molecular Neurogenetics is dedicated to research and advanced diagnostic activities to offer a service of excellence to patients, and make progress toward the elucidation of the causes and mechanisms leading to mitochondrial diseases. The Unit is also devoted to the molecular genetics diagnosis of NBIA and other neurodegenerative diseases.

Selected peer reviewed publications

From the first paper that she published in 1991 to present, Valeria Tiranti is Author of 81 publications indexed in PubMed and some chapters in scientific books. The h index, calculated using the ISI Web of Science database, is 33.

-Tiranti V, Zeviani M. Altered sulfide (H(2)S) metabolism in ethylmalonic encephalopathy. Cold Spring Harb Perspect Biol. 2013 Jan 1;5(1):a011437.

-Rodenburg RJ, Schoonderwoerd GC, Tiranti V, Taylor RW, Rötig A, Valente L, Invernizzi F, Chretien D, He L, Backx GP, Janssen KJ, Chinnery PF, Smeets HJ, de Coo IF, van den Heuvel LP. A multi-center comparison of diagnostic methods for the biochemical evaluation of suspected mitochondrial disorders. Mitochondrion. 2013 Jan;13(1):36-43.

-Indrieri A, van Rahden VA, Tiranti V, Morleo M, Iaconis D, Tammaro R, D'Amato I, Conte I, Maystadt I, Demuth S, Zvulunov A, Kutsche K, Zeviani M, Franco B.
Mutations in COX7B cause microphthalmia with linear skin lesions, an unconventional mitochondrial disease. Am J Hum Genet. 2012 Nov 2;91(5):942-9.

-Brunetti D, Dusi S, Morbin M, Uggetti A, Moda F, D'Amato I, Giordano C, d'Amati G, Cozzi A, Levi S, Hayflick S, Tiranti V. Pantothenate kinase-associated

neurodegeneration: altered mitochondria membrane potential and defective respiration in Pank2 knock-out mouse model. Hum Mol Genet. 2012; 21(24): 5294-305.

-Panteghini C, Zorzi G, Venco P, Dusi S, Reale C, Brunetti D, Chiapparini L, Zibordi F, Siegel B, Garavaglia B, Simonati A, Bertini E, Nardocci N, Tiranti V. C19orf12 and FA2H Mutations Are Rare in Italian Patients With Neurodegeneration With Brain Iron Accumulation. Semin Pediatr Neurol. 2012; 19(2): 75-81
-Invernizzi F, D'Amato I, Jensen PB, Ravaglia S, Zeviani M, Tiranti V. Microscale oxygraphy reveals OXPHOS impairment in MRC mutant cells. Mitochondrion. 2012; 12:328-35.
-Leoni V, Strittmatter L, Zorzi G, Zibordi F, Dusi S, Garavaglia B, Venco P, Caccia C, Souza AL, Deik A, Clish CB, Rimoldi M, Ciusani E, Bertini E, Nardocci N, Mootha VK, Tiranti V. Metabolic consequences of mitochondrial coenzyme A deficiency in patients with PANK2 mutations. Mol Genet Metab. 2012; 105: 463-71.
-Hartig MB, Iuso A, Haack T, Kmiec T, Jurkiewicz E, Heim K, Roeber S, Tarabin V, Dusi S, Krajewska-Walasek M, Jozwiak S, Hempel M, Winkelmann J, Elstner M, Oexle K, Klopstock T, Mueller-Felber W, Gasser T, Trenkwalder C, Tiranti V, Kretzschmar H, Schmitz G, Strom TM, Meitinger T, Prokisch H. Absence of an orphan mitochondrial protein, c19orf12, causes a distinct clinical subtype of neurodegeneration with brain iron accumulation. Am J Hum Genet. 2011; 89: 543-50:200-5.