PERSONAL INFORMATION

BERTINI ENRICO SILVIO

Children's Hospital Bambino Gesù, Piazza Sant' Onofrio, 4 - 00165 Roma

06 6859 2104

enricosilvio.bertini@opbg.net

Sex F | Date of birth 08/06/1950 | Nationality Italian

WORK EXPERIENCE

1981-1988 **Assistant in Neurology**
Bambino Gesu' Children's Hospital IRCCS, Rome

1988 -1996 **Associated in Neurology**
Bambino Gesu' Children's Hospital IRCCS, Rome

1997 to present **Head of the Unit of Unit of Neuromuscular and Neurodegenerative Disorders and the Laboratory of Molecular Medicine**
Bambino Gesu' Children's Hospital IRCCS, Rome

EDUCATION AND TRAINING

1975 **Degree MD - Medicine**
La Sapienza University, Rome, IT

1979 **Degree Res. Neurologist - Neurology**
La Sapienza University, Rome, IT

1985 **Degree Ass Res Scientist - Neuropathology**
S. Luc University, Bruxelles, BE

1987 - 1995 **Visiting Scientist – Neuropathology**
Columbia University, NY, USA

ADDITIONAL INFORMATION

Editorial Board: Neuromuscular Disorders, Neuropediatrics

Honorary Memberships:

2002 - 2008 Member Research Commitee European Neuromuscular Center
2002- present Member Research Committee Famiglie SMA
2005 - present Member Research Commitee European Leukodystrophy Association
2007 - 2009 Executive Board World Muscle Society
2009 - present Member SAB (Spinal Muscle Board) FSMA EUROPE

Scientific Societies Memberships
Child Neurology Society, World Muscle Society, ICNA, Associazione Italiana di Miologia

Research Interest
Diagnosis and follow-up of neuromuscular disorders; Trials in Spinal Muscular Atrophy; Collagen VI deficiency disorders; Congenital muscular dystrophies; Mitochondrial disorders; spastic paraplegia; Leukodystrophies; Congenital Ataxia.

Journal Reviews
Annals of Neurology, Neurology, Journal of Medical Genetics, Brain, American Journal of Human Genetics, European Journal of Human Genetics, Human Molecular Genetics, Brain Research, Neurogenetics,Clinical Genetics, Neuropediatrics, Neuromuscular Disorders, Genes, Journal of Inherited and Metabolic Disorders, Journal of Neurology, Journal of Neurological Sciences; Orphanet Journal of Rare Diseases.

Pubblications peer-reviewed (Index Medicus): 440

https://scholar.google.it/citations?user=qFhoz68AAAAJ&hl=en

H-index: 65

Last 5 pubblications 2014

1: Brea-Calvo G, Haack TB, Karall D, Ohtake A, Invernizzi F, Carrozzo R, Kremer L, Dusi S, Fauth C, Scholl-Bürgi S, Graf E, Ahting U, Resta N, Laforgia N, Verrigni D, Okazaki Y, Kohda M, Martinelli D, Freisinger P, Strom TM, Meitinger T, Lamperti C, Lacson A, Navas P, Mayr JA, Bertini E, Murayama K, Zeviani M, Prokisch H, Ghezzi D. COQ4 Mutations Cause a Broad Spectrum of Mitochondrial Disorders Associated with CoQ10 Deficiency. Am J Hum Genet. 2015 Feb 5;96(2):309-17.

2: Yuen M, Sandaradura SA, Dowling JJ, Kostyukova AS, Moroz N, Quinlan KG, Lehtokari VL, Ravenscroft G, Todd EJ, Ceyhan-Birsoy O, Gokhin DS, Maluenda J, Lek M, Nolent F, Pappas CT, Novak SM, D'Amico A, Malfatti E, Thomas BP, Gabriel SB, Gupta N, Daly MJ, Ilkovski B, Houweling PJ, Davidson AE, Swanson LC, Brownstein CA, Gupta VA, Medne L, Shannon P, Martin N, Bick DP, Flisberg A, Holmberg E, Van den Bergh P, Lapunzina P, Waddell LB, Sloboda DD, Bertini E, Chitayat D, Telfer WR, Laquerrière A, Gregorio CC, Ottenheijm CA, Bönnemann CG, Pelin K, Beggs AH, Hayashi YK, Romero NB, Laing NG, Nishino I, Wallgren-Pettersson C, Melki J, Fowler VM, MacArthur DG, North KN, Clarke NF. Leiomodin-3 dysfunction results in thin filament disorganization and nemaline myopathy. J Clin Invest. 2015 Jan;125(1):456-7.

3: Graziano A, Bianco F, D'Amico A, Moroni I, Messina S, Bruno C, Pegoraro E, Mora M, Astrea G, Magri F, Comi GP, Berardinelli A, Moggio M, Morandi L, Pini A, Petillo R, Tasca G, Monforte M, Minetti C, Mongini T, Ricci E, Gorni K, Battini R, Villanova M, Politano L, Gualandi F, Ferlini A, Muntoni F, Santorelli FM, Bertini E, Pane M, Mercuri E. Prevalence of congenital muscular dystrophy in Italy: A population study. Neurology. 2015 Mar 3;84(9):904-11.

4: Masotti A, Celluzzi A, Petrini S, Bertini E, Zanni G, Compagnucci C. Aged iPSCs display an uncommon mitochondrial appearance and fail to undergo in vitro neurogenesis. Aging (Albany NY). 2014 Dec;6(12):1094-108.

5: Kopajtich R, Nicholls TJ, Rorbach J, Metodiev MD, Freisinger P, Mandel H, Vanlander A, Ghezzi D, Carrozzo R, Taylor RW, Marquard K, Murayama K, Wieland T, Schwarzmayr T, Mayr JA, Pearce SF, Powell CA, Saada A, Ohtake A, Invernizzi F, Lamantea E, Sommerville EW, Pyle A, Chinnery PF, Crushell E, Okazaki Y, Kohda M, Kishita Y, Tokuzawa Y, Assouline Z, Rio M, Feillet F, Mousson de Camaret B, Chretien D, Munnich A, Menten B, Sante T, Smet J, Régal L, Lorber A, Khoury A, Zeviani M, Strom TM, Meitinger T, Bertini ES, Van Coster R, Klopstock T, Rötig A, Haack TB, Minczuk M, Prokisch H. Mutations in GTPBP3 cause a mitochondrial translation defect associated with hypertrophic cardiomyopathy, lactic acidosis, and encephalopathy. Am J Hum Genet. 2014 Dec 4;95(6):708-20.