PERSONAL INFORMATION

Enrico Bertini, MD

Ospedale Bambino Gesù IRCCS, Piazza Sant'Onofrio, 4 - 00165 Roma

CF BRTNCS50H08H501G (for Enrico Silvio Bertini)

WORK EXPERIENCE

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

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

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

EDUCATION AND TRAINING

Institution and place	Degree	Year(s)	Field of study
La Sapienza University, Rome, IT	MD	1975	Medicine
La Sapienza University, Rome, IT	Res. Neurologist	1979	Neurology
S. Luc University, Bruxelles, BE	Ass Res Scientist	1985	Neuropathology
Columbia University, NY, USA	Visiting Scientist	1987	Neuropathology
Columbia University, NY, USA	Visiting Scientist	1995	Neuropathology

ADDITIONAL INFORMATION

Teaching experience

Contract Professor in Neurogenetics at the School of Specialization in Medical Genetics, Catholic University of Rome, 1993-2019

Contract Professor of Neuromuscular Disorders at the University of Tor Vergata, Residency in Pediatrics, 2016-present

Editorial Board

Neuromuscular Disorders

Honorary Memberships:

2002 - 2008 Member Research Committee 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
2010 –present Member Research Committee MITOCON

Scientific Societies Memberships

Child Neurology Society, World Muscle Society, ICNA,
Associazione Italiana di Miologia , SIN (Società Italiana di Neurologia)

Participation to Clinical Trials

PTC Trial: *Phase 2b trial was a randomized, double-blind, placebo-controlled trial designed to evaluate the safety and efficacy of 48 weeks of ataluren therapy in Duchenne Muscular Dystrophy (2008-2010) PI*

TROPHOS: *A Phase II study to assess safety and efficacy of olesoxime (TRO19622) in 3-25 year old Spinal Muscular Atrophy (SMA) patients (2011-2012)*

GSK: *A Phase II, Double Blind, Exploratory, Parallel-group, Placebocontrolled Clinical Study to Assess Two Dosing Regimens of GSK2402968 for Efficacy, Safety, Tolerability and Pharmacokinetics in Ambulant Subjects With Duchenne Muscular Dystrophy*

BIOGEN-IONIS. Phase 2 NURTURE Study. Efficacy and Safety of Nusinersen in Genetically Diagnosed Infants with Presymptomatic Spinal Muscular Atrophy (SMA):

SPR1NT: A Global Study of a Single, One-Time Dose of AVXS-101 Delivered to Infants With Genetically Diagnosed and Pre-symptomatic Spinal Muscular Atrophy With Multiple Copies of SMN2-GB

Languages:

1. Italian; 2. English (speaks and writes); 3. Spanish (speaks and writes); 4. French (speaks and writes)

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, Nature Medicine, Mitochondrion, European Journal of Human Genetics, Human Molecular Genetics, Brain Research, Brain, 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): 776

H-index: 105 (Google Scholar)
H-index Scopus: 87
Last 5 pubblications 2021

1: Nuovo S, Baglioni V, De Mori R, Tardivo S, Caputi C, Ginevrino M, Micalizzi A, Masuelli L, Federici G, Casella A, Lorefice E, Anello D, Tolve M, Farini D,

Bertini E, Zanni G, Travaglini L, Vasco G, Sette C, Carducci C, Valente EM, Leuzzi V. Clinical variability at the mild end of BRAT1-related spectrum: Evidence from two families with genotype-phenotype discordance. Hum Mutat. 2021 Nov 8. doi: 10.1002/humu.24293. Epub ahead of print. PMID: 34747546.

2: Serpieri V, D'Abrusco F, Dempsey JC, Cheng YH, Arrigoni F, Baker J, Battini R, Bertini ES, Borgatti R, Christman AK, Curry C, D'Arrigo S, Fluss J, Freilinger M, Gana S, Ishak GE, Leuzzi V, Loucks H, Manti F, Mendelsohn N, Merlini L, Miller CV, Muhammad A, Nuovo S, Romaniello R, Schmidt W, Signorini S, Siliquini S, Szczałuba K, Vasco G, Wilson M, Zanni G, Boltshauser E, Doherty D, Valente EM; University of Washington Center for Mendelian Genomics (UW-CMG) group. SUFU haploinsufficiency causes a recognisable neurodevelopmental phenotype at the mild end of the Joubert syndrome spectrum. J Med Genet. 2021 Oct 21:jmedgenet-2021-108114. doi: 10.1136/jmedgenet-2021-108114. Epub ahead of print. PMID: 34675124.

3: D'Amico A, Longo A, Fattori F, Tosi M, Bosco L, Chiarini Testa MB, Paglietti G, Cherchi C, Carlesi A, Mizzoni I, Bertini E. Hepatobiliary disease in XLMTM: a common comorbidity with potential impact on treatment strategies. Orphanet J Rare Dis. 2021 Oct 12;16(1):425. doi: 10.1186/s13023-021-02055-1. PMID: 34641930; PMCID: PMC8513353.

4: Ardissone A, Bruno C, Diodato D, Donati A, Ghezzi D, Lamantea E, Lamperti C, Mancuso M, Martinelli D, Primiano G, Procopio E, Rubegni A, Santorelli F, Schiaffino MC, Servidei S, Tubili F, Bertini E, Moroni I. Clinical, imaging, biochemical and molecular features in Leigh syndrome: a study from the Italian network of mitochondrial diseases. Orphanet J Rare Dis. 2021 Oct 9;16(1):413. doi: 10.1186/s13023-021-02029-3. PMID: 34627336; PMCID: PMC8501644.

5: Helman G, Mendes MI, Nicita F, Darbelli L, Sherbini O, Moore T, Derksen A, Amy Pizzino, Carrozzo R, Torraco A, Catteruccia M, Aiello C, Goffrini P, Figuccia S, Smith DEC, Hadzsiev K, Hahn A, Biskup S, Brösse I, Kotzaeridou U, Gauck D, Grebe TA, Elmslie F, Stals K, Gupta R, Bertini E, Thiffault I, Taft RJ, Schiffmann R, Brandl U, Haack TB, Salomons GS, Simons C, Bernard G, van der Knaap MS, Vanderver A, Husain RA. Expanded phenotype of AARS1-related white matter disease. Genet Med. 2021 Aug 27. doi: 10.1038/s41436-021-01286-8. Epub ahead of print. PMID: 34446925.

The undersigned, aware that false declarations involve the application of the criminal sanctions provided for by art. 76 of Presidential Decree 445/2000, declares that the information contained in the following curriculum vitae, drawn up in European format, corresponds to the truth.
NB: pursuant to Law 196/03, the Entity / Company is authorized to process the personal data listed here for the exclusive purposes related to professional collaborations "

Rome, 05/05/2022

Signature_____  _____