

CURRICULUM VITAE

INFORMAZIONI PERSONALI

Cognome Nome	BRUNO CLAUDIO
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Nazionalità	Italiana
Data di nascita	27/12/1965

FORMAZIONE E STAGE

Tipologia	Research Fellow
Data	1996-1999
Sede	Dept. of Neurology, Columbia University, New York, USA

SPECIALIZZAZIONI

Tipologia	Pediatria
Data	1995
Sede	Università di Genova-Istituto Giannina Gaslini

ESPERIENZA LAVORATIVA

Data (da – a)	2001-oggi
Nome Istituzione	U.O. Malattie Muscolari e Neurodegenerative-Istituto G. Gaslini, Genova
Incarico ricoperto	Dirigente Medico 1° Livello

COORDINAMENTO GRUPPI DI LAVORO – GRUPPI DI RICERCA. NAZIONALI ED INTERNAZIONALI

Membro della Associazione Italiana Miologia (AIM) e della World Muscle Society (WMS).
Responsabile di finanziamenti da Fondazione Telethon e Istituzioni Italiane (Ministero della Salute).

ELENCO DELLE PUBBLICAZIONI SCIENTIFICHE PIÙ SIGNIFICATIVE	<u>Bruno C</u>, DiMauro S. Lipid Storage Myopathies. Curr Opin Neurol 2008; 21:601-606. <u>Bruno C</u>, Bertini E, Di Rocco M, et al. Clinical and genetic characterization of Chanarin-Dorfman syndrome. Biochem Biophys Res Commun 2008; 369:1125-1128. <u>Bruno C</u>, Cassandrini D, Assereto S, et al. Neuromuscular forms of glycogen branching enzyme deficiency. Acta Myologica 2007; 26:75-78. <u>Bruno C</u>, Cassandrini D, Martinuzzi A, et al. McArdle disease: the mutation spectrum of PYGM in a large Italian cohort. Hum Mutat 2006; 27(7):718. Biancheri R, Zara F, <u>Bruno C</u>, et al. Phenotypic characterization of hypomyelination and congenital cataract. Ann Neurol. 2007; 62:121-7. Zara F, Biancheri R, <u>Bruno C</u>, et al. Deficiency of hyccin, a newly identified membrane protein, causes hypomyelination and congenital cataract. Nat Genet 2006; 38:1111-3. <u>Bruno C</u>, van Diggelen OP, Cassandrini D, et al. Clinical and genetic heterogeneity of branching enzyme deficiency (glycogenosis type IV). Neurology 2004; 63:1053-108. <u>Bruno C</u>, Bertini E, Federico A, et al. Clinical and molecular findings in patients with giant axonal neuropathy (GAN). Neurology 2004; 62:13-16. <u>Bruno C</u>, Minetti C. Congenital myopathies. Curr Neurol Neurosci Rep 2004; 4:68-73. <u>Bruno C</u>, Santorelli FM, Assereto S, et al. Progressive exercise intolerance associated with a new muscle-restricted nonsense mutation (G142X) in the mitochondrial cytochrome b gene. Muscle Nerve 2003; 28:508-511. <u>Bruno C</u>, Sacco O, Santorelli FM, et al. Mitochondrial myopathy and respiratory failure associated with a new mutation in the mitochondrial transfer ribonucleic acid glutamic acid gene. J Child Neurol 2003; 18:300-303. <u>Bruno C</u>, Lanzillo R, Biedi C, et al. Two new mutations in the myophosphorylase gene in Italian patients with McArdle's disease. Neuromuscul Disord 2002; 12:498-500. <u>Bruno C</u>, Bado M, Minetti C, Cordone G, DiMauro S. Novel mutation in the CPT II gene in a child with periodic febrile myalgia and myoglobinuria. J Child Neurol 2000; 15:390-393. <u>Bruno C</u>, Bertini E, Santorelli FM, DiMauro S. HyperCKemia as the only sign of McArdle's disease in a child. J Child Neurol 2000; 15:137-138. <u>Bruno C</u>, Martinuzzi A, Tang Y, et al. A stop-codon mutation in the human mtDNA cytochrome c oxidase I gene disrupts the functional structure of complex IV. Am J Hum Genet 1999; 65:611-620. <u>Bruno C</u>, Kirby DM, Koga Y, et al. The mitochondrial DNA C3303T mutation can cause cardiomyopathy and/or skeletal myopathy. J Pediatr 1999; 135:197-202.
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ULTERIORI INFORMAZIONI	
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