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FORMAZIONE E STAGE

Tipologia		Research Fellow
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SPECIALIZZAZIONI

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ESPERIENZA LAVORATIVA

Data (da – a)		2001-oggi
Nome Istituzione		U.O. Malattie Muscolari e Neurodegenerative-Istituto G. Gaslini, Genova
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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		Bruno C, DiMauro S.Lipid Storage Myopathies. Curr Opin Neurol 2008; 21:601-606. Bruno C, Bertini E, Di Rocco M, et al. Clinical and genetic characterization of Chanarin-Dorfman syndrome. Biochem Biophys Res Commun 2008; 369:1125-1128. Bruno C, Cassandrini D, Assereto S, et al. Neuromuscular forms of glycogen branching enzyme deficiency. Acta Myologica 2007; 26:75-78. Bruno C, 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, Bruno C, et al. Phenotypic characterization of hypomyelination and congenital cataract. Ann Neurol. 2007, 62:121-7. Zara F, Biancheri R, Bruno C, et al. Deficiency of hyccin, a newly identified membrane protein, causes hypomyelination and congenital cataract. Nat Genet 2006, 38:1111-3. Bruno C, van Diggelen OP, Cassandrini D, et al. Clinical and genetic heterogeneity of branching enzyme deficiency (glycogenosis type IV). Neurology 2004; 63:1053-108. Bruno C, Bertini E, Federico A, et al. Clinical and molecular findings in patients with giant axonal neuropathy (GAN). Neurology 2004; 62:13-16. Bruno C, Minetti C. Congenital myopathies. Curr Neurol Neurosci Rep 2004; 4:68-73. Bruno C, 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. Bruno C, 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. Bruno C, 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. Bruno C, 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. Bruno C, 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. Bruno C, 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. Bruno C, Kirby DM, Koga Y, et al. The mitochondrial DNA C3303T mutation can cause cardiomyopathy and/or skeletal myopathy. J Pediatr 1999; 135:197-202.

ULTERIORI INFORMAZIONI		