



**LIST OF PUBLICATIONS 2004 ACKNOWLEDGING  
EUROBIOBANK**

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1. Angelini C. **Limb-girdle muscular dystrophies: heterogeneity of clinical phenotypes and pathogenetic mechanisms.** Acta Myologica. 13: 130-136; 2004.
2. Canki-Klain N, Milic A, Kovac B, Trlaja A, Grgicevic D, Zurak N, Fardeau M, Leturcq F, Kaplan JC, Urtizberea JA, Politano L, Piluso G, Feingold J. **Prevalence of the 550delA mutation in calpainopathy (LGMD 2A) in Croatia.** Am J Med Genet 2004 Mar 1;125(2):152-6.
3. Crimi M; Papadimitriou A; Galbiati S; Palamidou P; Fortunato F; Bordoni A; Papandreou U; Papadimitriou D; Hadjigeorgiou GM.; Drogari E; Bresolin N; Comi GP. **A New Mitochondrial DNA Mutation in ND3 Gene Causing Severe Leigh Syndrome with Early Lethality.** Pediatric Research.2004 May;55(5): 842-846
4. Di Blasi C, Moghadaszadeh B, Ciano C, Negri T, Giavazzi A, Cornelio F, Morandi L and Mora M. **Abnormal lysosomal and ubiquitin-proteasomal pathways in 19p13.3-linked distal myopathy.** Ann Neurol. 2004 Jul;56(1):133-8.
5. Fanin M, Fulizio L, Nascimbeni AC, Spinazzi M, Piluso G, Ventriglia VM, Ruzza G, Siciliano G, Trevisan CP, Politano L, Nigro V, Angelini C. **Molecular diagnosis of LGMD2A: mutation analysis or protein testing?** Human Mutation 2004 Jul;24(1): 52-62.
6. Horvath R, Lochmüller H, Hoeltzenbein M, Muller-Hocker J, Schoser B.G, Pongratz D, Jaksch M. **Spontaneous recovery of a childhood onset mitochondrial myopathy caused by a stop mutation in the mitochondrial cytochrome c oxydase III gene.** J Med Genet.2004 ; 41:75
7. Jaeger C, Galea D, Angelini C, Bignami F, Cohen O, Di Donato JH, Guitard-Arnau C, Izquierdo Martinez M, Karcagi V, Laurent JC, Meznaric-Paz M, Salama F, Verellen-Dumoulin C, Lochmüller H. **EuroBioBank (EBB): European Network of DNA, Cell and Tissue Banks for Rare Diseases.** European Journal of Human Genetics, 2004, 12, Suppl. 1: 330
8. Politano L. **Eurobiobank, a European Network of DNA, Cell and Tissue Banks for Rare Diseases.** Acta Myologica, 2004, vol 23 (1): 76
9. Scarlato M, Carlo Previtalli S, Carpo M, Pareyson D, Briani C, Del Bo R, Nobile-Orazio E, Quattrini A and Comi GP. **Polyneuropathy in POEMS syndrome: role of angiogenic factors in the pathogenesis.** Brain 2005; 128(8):1911-1920
10. Schoser BG, Kress W, Walter M, Halliger-Keller B, Lochmüller H, Ricker K. **Homozygosity for CCTG mutation in myotonic dystrophy type 2.** Brain 2004, 127: 1868-1877