



MASTER PEDIATRIC ENDOCRINOLOGY AND DIABETES

Amelia Morrone list of publications

Cavicchi C, Chilleri C, Fioravanti A, Ferri L, Ripandelli F, Costa C, Calabresi P, Prontera P, Pochiero F, Pasquini E, Funghini S, la Marca G, Donati MA, **Morrone A**. Late-Onset N-Acetylglutamate Synthase Deficiency: Report of a Paradigmatic Adult Case Presenting with Headaches and Review of the Literature. *Int J Mol Sci*. 2018 Jan 24;19(2). pii: E345. doi: 10.3390/ijms19020345. [[ABSTRACT](#)] [[FULL TEXT](#)]

Lora V, De Felice C, Cota C, Graceffa D, **Morrone A**, Bonifati C. A case of juvenile pityriasis rubra pilaris type III successfully treated with etanercept. *Dermatol Ther*. 2017 Nov 28. doi: 10.1111/dth.12577. [[ABSTRACT](#)]

Ardissino G, Perrone M, Tel F, Testa S, **Morrone A**, Possenti I, Tagliaferri F, Dilena R, Menni F. Late Onset Cobalamin Disorder and Hemolytic Uremic Syndrome: A Rare Cause of Nephrotic Syndrome. *Case Rep Pediatr*. 2017;2017:2794060. doi:10.1155/2017/2794060. [[ABSTRACT](#)] [[FULL TEXT](#)] [[PDF](#)]

Deodato F, Procopio E, Rampazzo A, Taurisano R, Donati MA, Dionisi-Vici C, Caciotti A, **Morrone A**, Scarpa M. The treatment of juvenile/adult GM1-gangliosidosis with Miglustat may reverse disease progression. *Metab Brain Dis*. 2017 Oct;32(5):1529-1536. doi: 10.1007/s11011-017-0044-y. [[ABSTRACT](#)]

Bacci GM, Donati MA, Pasquini E, Munier F, Cavicchi C, **Morrone A**, Sodi A, Murro V, Garcia Segarra N, Defilippi C, Bussolin L, Caputo R. Optical coherence tomography morphology and evolution in cbIC disease-related maculopathy in a case series of very young patients. *Acta Ophthalmol*. 2017 Dec;95(8):e776-e782. doi:10.1111/aos.13441. [[ABSTRACT](#)]

Ferri L, Covello G, Caciotti A, Guerrini R, Denti MA, **Morrone A**. Double-target Antisense U1snRNAs Correct Mis-splicing Due to c.639+861C>T and c.639+919G>A GLA Deep Intronic Mutations. *Mol Ther Nucleic Acids*. 2016 Oct 25;5(10):e380. doi:10.1038/mtna.2016.88. [[ABSTRACT](#)] [[FULL TEXT](#)] [[PDF](#)]

Ferri L, Dionisi-Vici C, Taurisano R, Vaz FM, Guerrini R, **Morrone A**. When silence is noise: infantile-onset Barth syndrome caused by a synonymous substitution affecting TAZ gene transcription. *Clin Genet*. 2016 Nov;90(5):461-465. doi: 10.1111/cge.12756. [[ABSTRACT](#)]

Front S, Biela-Banaś A, Burda P, Ballhausen D, Higaki K, Caciotti A, **Morrone A**, Charollais-Thoenig J, Gallienne E, Demotz S, Martin OR. (5aR)-5a-C-Pentyl-4-epi-isofagomine: A powerful inhibitor of lysosomal β -galactosidase and a remarkable chaperone for mutations associated with GM1-gangliosidosis and Morquio disease type B. *Eur J Med Chem*. 2017 Jan 27;126:160-170. doi: 10.1016/j.ejmech.2016.09.095. [[ABSTRACT](#)]

Barba C, Darra F, Cusmai R, Procopio E, Dionisi Vici C, Keldermans L, Vuillaumier-Barrot S, Lefeber DJ, Guerrini R; CDG Group. Congenital disorders of glycosylation presenting as epileptic encephalopathy with migrating partial seizures in infancy. *Dev Med Child Neurol*. 2016 Oct;58(10):1085-91. doi:10.1111/dmcn.13141. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

Verrecchia E, Zampetti A, Antuzzi D, Ricci R, Ferri L, **Morrone A**, Feliciani C, Dagna L, Manna R. The impact of fever/hyperthermia in the diagnosis of Fabry: A retrospective analysis. *Eur J Intern Med*. 2016 Jul;32:26-30. doi:10.1016/j.ejim.2016.03.015. [\[ABSTRACT\]](#)

Tonin R, Caciotti A, Funghini S, Pasquini E, Mooney SD, Cai B, Proncopio E, Donati MA, Baronio F, Bettocchi I, Cassio A, Biasucci G, Bordugo A, la Marca G, Guerrini R, **Morrone A**. Clinical relevance of short-chain acyl-CoA dehydrogenase (SCAD) deficiency: Exploring the role of new variants including the first SCAD-disease-causing allele carrying a synonymous mutation. *BBA Clin*. 2016 Mar 10;5:114-9. doi: 10.1016/j.bbacli.2016.03.004. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

Pieruzzi F, Pieroni M, Zachara E, Marziliano N, **Morrone A**, Cecchi F. [Heart involvement in Anderson-Fabry disease: Italian recommendations for diagnostic, follow-up and therapeutic management]. *G Ital Cardiol (Rome)*. 2015 Nov;16(11):630-8. doi: 10.1714/2066.22434. [\[ABSTRACT\]](#)

Ferri L, Cavicchi C, Fiumara A, Parini R, Guerrini R, **Morrone A**. Pitfalls in the detection of gross gene rearrangements using MLPA in Fabry disease. *Clin Chim Acta*. 2016 Jan 15;452:82-6. doi: 10.1016/j.cca.2015.10.027. [\[ABSTRACT\]](#)

Parmeggiani C, Catarzi S, Matassini C, D'Adamio G, **Morrone A**, Goti A, Paoli P, Cardona F. Human Acid β -Glucosidase Inhibition by Carbohydrate Derived Iminosugars: Towards New Pharmacological Chaperones for Gaucher Disease. *Chembiochem*. 2015 Sep 21;16(14):2054-64. doi: 10.1002/cbic.201500292. [\[ABSTRACT\]](#)

Romani I, Borsini W, Nencini P, **Morrone A**, Ferri L, Frusconi S, Donadio VA, Liguori R, Donati MA, Falconi S, Pracucci G, Inzitari D. De novo Diagnosis of Fabry Disease among Italian Adults with Acute Ischemic Stroke or Transient Ischemic Attack. *J Stroke Cerebrovasc Dis*. 2015 Nov;24(11):2588-95. doi:10.1016/j.jstrokecerebrovasdis.2015.07.012. [\[ABSTRACT\]](#)

Mignani R, Gallieni M, Feriozzi S, Pisani A, Marziliano N, **Morrone A**. [The nephropathy in the Anderson-Fabry disease: new recommendations for the diagnosis, the follow-up and the therapy]. *G Ital Nefrol*. 2015 Jul-Aug;32(4). pii:gin/32.4.11. [\[ABSTRACT\]](#)

Tonin R, Caciotti A, Funghini S, la Marca G, Pasquini E, Cayton E, Mooney SD, Guerrini R, **Morrone A**. Biotinidase deficiency due to a de novo mutation or gonadal mosaicism in a first child. *Clin Chim Acta*. 2015 May 20;445:70-2. doi:10.1016/j.cca.2015.03.010. [\[ABSTRACT\]](#)

Ferri L, Donati MA, Funghini S, Cavicchi C, Pensato V, Gellera C, Natacci F, Spaccini L, Gasperini S, Vaz FM, Cooper DN, Guerrini R, **Morrone A**. Intra-individual plasticity of the TAZ gene leading to different heritable mutations in siblings with Barth syndrome. *Eur J Hum Genet*. 2015 Dec;23(12):1708-12. doi: 10.1038/ejhg.2015.50. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

Parrini E, Mei D, Pisanti MA, Catarzi S, Pucatti D, Bianchini C, Mascalchi M, Bertini E, **Morrone A**, Cavaliere ML, Guerrini R. Familial periventricular nodular heterotopia, epilepsy and Melnick-Needles Syndrome caused by a single FLNA mutation with combined gain-of-function and loss-of-function effects. *J Med Genet*. 2015 Jun;52(6):405-12. doi: 10.1136/jmedgenet-2014-102959. [\[ABSTRACT\]](#)

Sacchini M, Procopio E, Pasquini E, Pochiero F, Ombrone D, LaMarca G, Catarzi S, **Morrone A**, Donati MA. Alpha Glucosidase Assay on Dried Blood Spot in the Early Diagnosis of Infantile Pompe Disease. *J Neuromuscul Dis*. 2015;2(s1):S53. [\[ABSTRACT\]](#)

Caciotti A, Tonin R, Rigoldi M, Ferri L, Catarzi S, Cavicchi C, Procopio E, Donati MA, Ficcadenti A, Fiumara A, Barone R, Garavelli L, Rocco MD, Filocamo M, Antuzzi D, Scarpa M, Mooney SD, Li B, Skouma A, Bianca S, Concolino D, Casalone R,

Monti E, Pantaleo M, Giglio S, Guerrini R, Parini R, **Morrone A**. Optimizing the molecular diagnosis of GALNS: novel methods to define and characterize Morquio-A syndrome-associated mutations. *Hum Mutat*. 2015 Mar;36(3):357-68. doi:10.1002/humu.22751. [\[ABSTRACT\]](#)

Morrone A, Caciotti A, Atwood R, Davidson K, Du C, Francis-Lyon P, Harmatz P, Mealiffe M, Mooney S, Oron TR, Ryles A, Zawadzki KA, Miller N. Morquio A syndrome-associated mutations: a review of alterations in the GALNS gene and a new locus-specific database. *Hum Mutat*. 2014 Nov;35(11):1271-9. doi:10.1002/humu.22635. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

Cavicchi C, Donati M, Parini R, Rigoldi M, Bernardi M, Orfei F, Gentiloni Silveri N, Colasante A, Funghini S, Catarzi S, Pasquini E, la Marca G, Mooney S, Guerrini R, **Morrone A**. Sudden unexpected fatal encephalopathy in adults with OTC gene mutations-Clues for early diagnosis and timely treatment. *Orphanet J Rare Dis*. 2014 Jul 16;9:105. doi: 10.1186/s13023-014-0105-9. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

Morrone A, Tylee KL, Al-Sayed M, Brusius-Facchin AC, Caciotti A, Church HJ, Coll MJ, Davidson K, Fietz MJ, Gort L, Hegde M, Kubaski F, Lacerda L, Laranjeira F, Leistner-Segal S, Mooney S, Pajares S, Pollard L, Ribeiro I, Wang RY, Miller N. Molecular testing of 163 patients with Morquio A (Mucopolysaccharidosis IVA) identifies 39 novel GALNS mutations. *Mol Genet Metab*. 2014 Jun;112(2):160-70. doi: 10.1016/j.ymgme.2014.03.004. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

Catarzi S, Caciotti A, Thusberg J, Tonin R, Malvagia S, la Marca G, Pasquini E, Cavicchi C, Ferri L, Donati MA, Baronio F, Guerrini R, Mooney SD, **Morrone A**. Medium-chain acyl-CoA deficiency: outlines from newborn screening, in silico predictions, and molecular studies. *ScientificWorldJournal*. 2013 Oct 31;2013:625824. doi: 10.1155/2013/625824. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

Ferri L, Funghini S, Fioravanti A, Biondi EG, la Marca G, Guerrini R, Donati MA, **Morrone A**. Aminoacylase I deficiency due to ACY1 mRNA exon skipping. *Clin Genet*. 2014 Oct;86(4):367-72. doi: 10.1111/cge.12297. [\[ABSTRACT\]](#)

Rigoldi M, Concolino D, **Morrone A**, Pieruzzi F, Ravaglia R, Furlan F, Santus F, Strisciuglio P, Torti G, Parini R. Intrafamilial phenotypic variability in four families with Anderson-Fabry disease. *Clin Genet*. 2014 Sep;86(3):258-63. doi: 10.1111/cge.12261. [\[ABSTRACT\]](#)

Caciotti A, Catarzi S, Tonin R, Lugli L, Perez CR, Michelakakis H, Mavridou I, Donati MA, Guerrini R, d'Azzo A, **Morrone A**. Galactosialidosis: review and analysis of CTSA gene mutations. *Orphanet J Rare Dis*. 2013 Aug 2;8:114. doi:10.1186/1750-1172-8-114. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

Tomberli B, Cecchi F, Sciagrà R, Berti V, Lisi F, Torricelli F, **Morrone A**, Castelli G, Yacoub MH, Olivotto I. Coronary microvascular dysfunction is an early feature of cardiac involvement in patients with Anderson-Fabry disease. *Eur J Heart Fail*. 2013 Dec;15(12):1363-73. doi: 10.1093/eurjhf/hft104. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

Tummolo A, Favia V, Bellantuono R, Bellino V, Ranieri A, **Morrone A**, De Palo T, Papadia F. Successful early management of a female patient with a metabolic stroke due to ornithine transcarbamylase deficiency. *Pediatr Emerg Care*. 2013 May;29(5):656-8. doi: 10.1097/PEC.0b013e31828ec2b9. [\[ABSTRACT\]](#)

Spada M, Enea A, **Morrone A**, Fea A, Porta F. Cornea verticillata and Fabry disease. *J Pediatr*. 2013 Aug;163(2):609. doi: 10.1016/j.jpeds.2013.03.013. [\[ABSTRACT\]](#)

Ferri L, Caciotti A, Cavicchi C, Rigoldi M, Parini R, Caserta M, Chibbaro G, Gasperini S, Procopio E, Donati MA, Guerrini R, **Morrone A**. Integration of PCR-Sequencing Analysis with Multiplex Ligation-Dependent Probe Amplification for Diagnosis of Hereditary Fructose Intolerance. *JIMD Rep*. 2012;6:31-7. doi:10.1007/8904_2012_125. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

Ferri L, Donati MA, Funghini S, Malvagia S, Catarzi S, Lugli L, Ragni L, Bertini E, Vaz FM, Cooper DN, Guerrini R, **Morrone A**. New clinical and molecular insights on Barth syndrome. *Orphanet J Rare Dis*. 2013 Feb 14;8:27. doi:10.1186/1750-1172-8-27. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

Zampetti A, Fania L, Antuzzi D, Giurdanella F, Gnarra M, Bertola F, Lualdi S, Filocamo M, **Morrone A**, Feliciani C. Mutation identification of Fabry disease in families with other lysosomal storage disorders. *Clin Genet*. 2013 Sep;84(3):281-5. doi: 10.1111/cge.12071. [\[ABSTRACT\]](#)

Lovera C, Porta F, Caciotti A, Catarzi S, Cassanello M, Caruso U, Gallina MR, **Morrone A**, Spada M. Sudden unexpected infant death (SUDI) in a newborn due to medium chain acyl CoA dehydrogenase (MCAD) deficiency with an unusual severe genotype. *Ital J Pediatr*. 2012 Oct 24;38:59. doi: 10.1186/1824-7288-38-59. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

Zampetti A, Orteu CH, Antuzzi D, Bongiorno MR, Manco S, Gnarra M, **Morrone A**, Cardinali G, Kovacs D, Aspite N, Linder D, Parini R, Feliciani C; Interdisciplinary Study Group on Fabry Disease (ISGF). Angiokeratoma: decision-making aid for the diagnosis of Fabry disease. *Br J Dermatol*. 2012 Apr;166(4):712-20. doi: 10.1111/j.1365-2133.2012.10742.x. [\[ABSTRACT\]](#)

Catarzi S, Giunti L, Papadia F, Gabrielli O, Guerrini R, Donati MA, Genuardi M, **Morrone A**. Morquio A syndrome due to maternal uniparental isodisomy of the telomeric end of chromosome 16. *Mol Genet Metab*. 2012 Mar;105(3):438-42. doi:10.1016/j.ymgme.2011.11.196. [\[ABSTRACT\]](#)

Funghini S, Thusberg J, Spada M, Gasperini S, Parini R, Ventura L, Meli C, De Cosmo L, Sibilio M, Mooney SD, Guerrini R, Donati MA, **Morrone A**. Carbamoyl phosphate synthetase 1 deficiency in Italy: clinical and genetic findings in a heterogeneous cohort. *Gene*. 2012 Feb 10;493(2):228-34. doi:10.1016/j.gene.2011.11.052. [\[ABSTRACT\]](#)

Ferri L, Guido C, la Marca G, Malvagia S, Cavicchi C, Fiumara A, Barone R, Parini R, Antuzzi D, Feliciani C, Zampetti A, Manna R, Giglio S, Della Valle CM, Wu X, Valenzano KJ, Benjamin R, Donati MA, Guerrini R, Genuardi M, **Morrone A**. Fabry disease: polymorphic haplotypes and a novel missense mutation in the GLA gene. *Clin Genet*. 2012 Mar;81(3):224-33. doi: 10.1111/j.1399-0004.2011.01689.x. [\[ABSTRACT\]](#)

Filocamo M, **Morrone A**. Lysosomal storage disorders: molecular basis and laboratory testing. *Hum Genomics*. 2011 Mar;5(3):156-69. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

Caciotti A, Garman SC, Rivera-Colón Y, Procopio E, Catarzi S, Ferri L, Guido C, Martelli P, Parini R, Antuzzi D, Battini R, Sibilio M, Simonati A, Fontana E, Salviati A, Akinci G, Cereda C, Dionisi-Vici C, Deodato F, d'Amico A, d'Azzo A, Bertini E, Filocamo M, Scarpa M, di Rocco M, Tifft CJ, Ciani F, Gasperini S, Pasquini E, Guerrini R, Donati MA, **Morrone A**. GM1 gangliosidosis and Morquio B disease: an update on genetic alterations and clinical findings. *Biochim Biophys Acta*. 2011 Jul;1812(7):782-90. doi: 10.1016/j.bbadis.2011.03.018. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

la Marca G, Malvagia S, Pasquini E, Cavicchi C, **Morrone A**, Ciani F, Funghini S, Villanelli F, Zammarchi E, Guerrini R. Newborn Screening for Tyrosinemia Type I: Further Evidence that Succinylacetone Determination on Blood Spot Is Essential. *JIMD Rep*. 2011;1:107-9. doi: 10.1007/8904_2011_24. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

Filoni C, Caciotti A, Carraresi L, Cavicchi C, Parini R, Antuzzi D, Zampetti A, Feriozzi S, Poisetti P, Garman SC, Guerrini R, Zammarchi E, Donati MA, **Morrone A**. Functional studies of new GLA gene mutations leading to conformational Fabry disease. *Biochim Biophys Acta*. 2010 Feb;1802(2):247-52. doi:10.1016/j.bbadis.2009.11.003. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

Houtkooper RH, Turkenburg M, Poll-The BT, Karall D, Pérez-Cerdá C, **Morrone A**, Malvagia S, Wanders RJ, Kulik W, Vaz FM. The enigmatic role of tafazzin in cardiolipin metabolism. *Biochim Biophys Acta*. 2009 Oct;1788(10):2003-14. doi: 10.1016/j.bbamem.2009.07.009. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

Filippi L, Gozzini E, Cavicchi C, Morrone A, Fiorini P, Donzelli G, Malvagia S, la Marca G. Insulin-resistant hyperglycaemia complicating neonatal onset of methylmalonic and propionic acidemias. *J Inher Metab Dis*. 2009 Dec;32 Suppl 1:S179-86. doi: 10.1007/s10545-009-1141-9. [\[ABSTRACT\]](#)

Caciotti A, Di Rocco M, Filocamo M, Grossi S, Traverso F, d'Azzo A, Cavicchi C, Messeri A, Guerrini R, Zammarchi E, Donati MA, Morrone A. Type II sialidosis: review of the clinical spectrum and identification of a new splicing defect with chitotriosidase assessment in two patients. *J Neurol*. 2009 Nov;256(11):1911-5. doi: 10.1007/s00415-009-5213-4. [\[ABSTRACT\]](#)

Mignani R, Morrone A. Is standard GLA gene mutation analysis definitive for the diagnosis of Fabry disease? *Kidney Int*. 2009 May;75(10):1115-6; author reply 1115. doi: 10.1038/ki.2009.28. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

Cavicchi C, Malvagia S, la Marca G, Gasperini S, Donati MA, Zammarchi E, Guerrini R, Morrone A, Pasquini E. Hypocitrullinemia in expanded newborn screening by LC-MS/MS is not a reliable marker for ornithine transcarbamylase deficiency. *J Pharm Biomed Anal*. 2009 Jul 12;49(5):1292-5. doi:10.1016/j.jpba.2009.03.001. [\[ABSTRACT\]](#)

Carraresi L, Parini R, Filoni C, Caciotti A, Sersale G, Tomatsu S, Orlando C, Zammarchi E, Guerrini R, Donati MA, Morrone A. GALNS gene expression profiling in Morquio A patients' fibroblasts. *Clin Chim Acta*. 2008 Nov;397(1-2):72-6. doi:10.1016/j.cca.2008.07.021. [\[ABSTRACT\]](#)

Caciotti A, Donati MA, d'Azzo A, Salvioli R, Guerrini R, Zammarchi E, Morrone A. The potential action of galactose as a "chemical chaperone": increase of beta galactosidase activity in fibroblasts from an adult GM1-gangliosidosis patient. *Eur J Paediatr Neurol*. 2009 Mar;13(2):160-4. doi:10.1016/j.ejpn.2008.03.004. [\[ABSTRACT\]](#)

Filoni C, Caciotti A, Carraresi L, Donati MA, Mignani R, Parini R, Filocamo M, Soliani F, Simi L, Guerrini R, Zammarchi E, Morrone A. Unbalanced GLA mRNAs ratio quantified by real-time PCR in Fabry patients' fibroblasts results in Fabry disease. *Eur J Hum Genet*. 2008 Nov;16(11):1311-7. doi: 10.1038/ejhg.2008.109. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

Parini R, Rigoldi M, Santus F, Furlan F, De Lorenzo P, Valsecchi G, Concolino D, Strisciuglio P, Feriozzi S, Di Vito R, Ravaglia R, Ricci R, Morrone A. Enzyme replacement therapy with agalsidase alfa in a cohort of Italian patients with Anderson-Fabry disease: testing the effects with the Mainz Severity Score Index. *Clin Genet*. 2008 Sep;74(3):260-6. doi:10.1111/j.1399-0004.2008.01012.x. [\[ABSTRACT\]](#)

Caciotti A, Donati MA, Adami A, Guerrini R, Zammarchi E, Morrone A. Different genotypes in a large Italian family with recurrent hereditary fructose intolerance. *Eur J Gastroenterol Hepatol*. 2008 Feb;20(2):118-21. doi:10.1097/MEG.0b013e3282f172e6. [\[ABSTRACT\]](#)

Malvagia S, Papi L, Morrone A, Donati MA, Ciani F, Pasquini E, la Marca G, Scholte HR, Genuardi M, Zammarchi E. Fatal malonyl CoA decarboxylase deficiency due to maternal uniparental isodisomy of the telomeric end of chromosome 16. *Ann Hum Genet*. 2007 Nov;71(Pt 6):705-12. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

Caciotti A, Donati MA, Procopio E, Filocamo M, Kleijer W, Wuyts W, Blaumeiser B, d'Azzo A, Simi L, Orlando C, McKenzie F, Fiumara A, Zammarchi E, Morrone A. GM1 gangliosidosis: molecular analysis of nine patients and development of an RT-PCR assay for GLB1 gene expression profiling. *Hum Mutat*. 2007 Feb;28(2):204. [\[ABSTRACT\]](#) [\[PDF\]](#)

Donati MA, Malvagia S, Pasquini E, Morrone A, La Marca G, Garavaglia B, Toniolo D, Zammarchi E. Barth syndrome presenting with acute metabolic decompensation in the neonatal period. *J Inher Metab Dis*. 2006 Oct;29(5):684. [\[ABSTRACT\]](#)

Lee BY, Han JA, Im JS, Morrone A, Johung K, Goodwin EC, Kleijer WJ, DiMaio D, Hwang ES. Senescence-associated beta-galactosidase is lysosomal beta-galactosidase. *Aging Cell*. 2006 Apr;5(2):187-95. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

- Burlina AB, Peduto A, Di Palma A, Bellizzi A, Sperli D, Morrone A, Burlina AP. An unusual clinical and biochemical presentation of ornithine transcarbamylase deficiency in a male patient. *J Inherit Metab Dis*. 2006 Feb;29(1):179-81. [\[ABSTRACT\]](#)
- Sinigerska I, Chandler D, Vaghjiani V, Hassanova I, Gooding R, Morrone A, Kremensky I, Kalaydjieva L. Founder mutation causing infantile GM1-gangliosidosis in the Gypsy population. *Mol Genet Metab*. 2006 May;88(1):93-5. [\[ABSTRACT\]](#)
- Cavicchi C, Donati MA, Funghini S, la Marca G, Malvagias S, Ciani F, Poggi GM, Pasquini E, Zammarchi E, Morrone A. Genetic and biochemical approach to early prenatal diagnosis in a family with mutant methylmalonic aciduria. *Clin Genet*. 2006 Jan;69(1):72-6. [\[ABSTRACT\]](#)
- Cavicchi C, Donati MA, Pasquini E, Poggi GM, Dionisi-Vici C, Parini R, Zammarchi E, Morrone A. Mutational spectrum in ten Italian patients affected by methylmalonyl-CoA mutase deficiency. *J Inherit Metab Dis*. 2005;28(6):1175-8. [\[ABSTRACT\]](#)
- Caciotti A, Donati MA, Bardelli T, d'Azzo A, Massai G, Luciani L, Zammarchi E, Morrone A. Primary and secondary elastin-binding protein defect leads to impaired elastogenesis in fibroblasts from GM1-gangliosidosis patients. *Am J Pathol*. 2005 Dec;167(6):1689-98. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)
- Malvagias S, Morrone A, Pasquini E, Funghini S, la Marca G, Zammarchi E, Donati MA. First prenatal molecular diagnosis in a family with holocarboxylase synthetase deficiency. *Prenat Diagn*. 2005 Dec;25(12):1117-9. [\[ABSTRACT\]](#)
- Funghini S, Morrone A, Pasquini E, Zammarchi E, Donati MA. Successful prenatal molecular diagnosis of carbamyl-phosphate synthetase I deficiency in two at-risk pregnancies. *J Inherit Metab Dis*. 2005;28(5):801-2. [\[ABSTRACT\]](#)
- Drousiotou A, Georgiou T, Drousiotou A, Campos Y, Caciotti A, Sztriha L, Gururaj A, Ozand P, Zammarchi E, Morrone A, d'Azzo A. Gene symbol: GLB1. Disease: GM1 gangliosidosis infantile. *Hum Genet*. 2005 May;116(6):542. [\[ABSTRACT\]](#)
- Drousiotou A, Georgiou T, Drousiotou A, Campos Y, Caciotti A, Sztriha L, Gururaj A, Ozand P, Zammarchi E, Morrone A, d'Azzo A. Gene symbol: GLB1. Disease: GM1 gangliosidosis infantile. *Hum Genet*. 2005 May;116(6):542. [\[ABSTRACT\]](#)
- Drousiotou A, Georgiou T, Drousiotou A, Campos Y, Caciotti A, Sztriha L, Gururaj A, Ozand P, Zammarchi E, Morrone A, d'Azzo A. Gene symbol: GLB1. Disease: GM1 gangliosidosis infantile. *Hum Genet*. 2005 May;116(6):534. [\[ABSTRACT\]](#)
- Georgiou T, Stylianidou G, Anastasiadou V, Caciotti A, Campos Y, Zammarchi E, Morrone A, D'azzo A, Drousiotou A. The Arg482His mutation in the beta-galactosidase gene is responsible for a high frequency of GM1 gangliosidosis carriers in a Cypriot village. *Genet Test*. 2005 Summer;9(2):126-32. [\[ABSTRACT\]](#)
- Caciotti A, Donati MA, Boneh A, d'Azzo A, Federico A, Parini R, Antuzzi D, Bardelli T, Nosi D, Kimonis V, Zammarchi E, Morrone A. Role of beta-galactosidase and elastin binding protein in lysosomal and nonlysosomal complexes of patients with GM1-gangliosidosis. *Hum Mutat*. 2005 Mar;25(3):285-92. [\[ABSTRACT\]](#)
- Ricci R, Castorina M, Di Lillo M, Antuzzi D, Frustaci A, Parini R, Menni F, Furlan F, Burlina A, Burlina A, Catuogno S, Gabrielli O, Burattini I, Borsini W, Buchner S, Ferriozzi S, Spisni C, De Vito R, Di Rocco M, Aricò M, Pistone G, Bongiorno AM, Morrone A, Cavicchi C, Zammarchi E. [Fabry disease in Italy: first epidemiologic and collaborative study]. *Ann Ital Med Int*. 2004 Oct-Dec;19(4):269-75. [\[ABSTRACT\]](#)
- Georgiou T, Drousiotou A, Campos Y, Caciotti A, Sztriha L, Gururaj A, Ozand P, Zammarchi E, Morrone A, D'Azzo A. Four novel mutations in patients from the Middle East with the infantile form of GM1-gangliosidosis. *Hum Mutat*. 2004 Oct;24(4):352. [\[ABSTRACT\]](#) [\[PDF\]](#)

Malvagia S, Morrone A, Caciotti A, Bardelli T, d'Azzo A, Ancora G, Zammarchi E, Donati MA. New mutations in the PPBG gene lead to loss of PPCA protein which affects the level of the beta-galactosidase/neuraminidase complex and the EBP-receptor. *Mol Genet Metab*. 2004 May;82(1):48-55. [\[ABSTRACT\]](#)

la Marca G, Malvagia S, Donati MA, **Morrone A**, Pasquini E, Zammarchi E. Rapid diagnosis of medium chain Acyl Co-A dehydrogenase (MCAD) deficiency in a newborn by liquid chromatography/tandem mass spectrometry. *Rapid Commun Mass Spectrom*. 2003;17(23):2688-92. [\[ABSTRACT\]](#)

Funghini S, Donati MA, Pasquini E, Zammarchi E, **Morrone A**. Structural organization of the human carbamyl phosphate synthetase I gene (CPS1) and identification of two novel genetic lesions. *Hum Mutat*. 2003 Oct;22(4):340-1. [\[ABSTRACT\]](#) [\[PDF\]](#)

Morrone A, Cavicchi C, Bardelli T, Antuzzi D, Parini R, Di Rocco M, Feriozzi S, Gabrielli O, Barone R, Pistone G, Spisni C, Ricci R, Zammarchi E. Fabry disease: molecular studies in Italian patients and X inactivation analysis in manifesting carriers. *J Med Genet*. 2003 Aug;40(8):e103. [\[ABSTRACT\]](#) [\[PDF\]](#)

Malvagia S, Poggi GM, Pasquini E, Donati MA, Pela I, **Morrone A**, Zammarchi E. The de novo Q167K mutation in the POU1F1 gene leads to combined pituitary hormone deficiency in an Italian patient. *Pediatr Res*. 2003 Nov;54(5):635-40. [\[ABSTRACT\]](#)

Caciotti A, Bardelli T, Cunningham J, D'Azzo A, Zammarchi E, **Morrone A**. Modulating action of the new polymorphism L436F detected in the GLB1 gene of a type-II GM1 gangliosidosis patient. *Hum Genet*. 2003 Jul;113(1):44-50. [\[ABSTRACT\]](#)

Caciotti A, **Morrone A**, Domenici R, Donati MA, Zammarchi E. Severe prognosis in a large family with hypokalemic periodic paralysis. *Muscle Nerve*. 2003 Feb;27(2):165-9. [\[ABSTRACT\]](#)

Bardelli T, Donati MA, Gasperini S, Ciani F, Belli F, Blau N, **Morrone A**, Zammarchi E. Two novel genetic lesions and a common BH4-responsive mutation of the PAH gene in Italian patients with hyperphenylalaninemia. *Mol Genet Metab*. 2002 Nov;77(3):260-6. [\[ABSTRACT\]](#)

Funghini S, Donati MA, Pasquini E, Gasperini S, Ciani F, **Morrone A**, Zammarchi E. Two new mutations in children affected by partial biotinidase deficiency ascertained by newborn screening. *J Inher Metab Dis*. 2002 Aug;25(4):328-30. [\[ABSTRACT\]](#)

Morrone A, Malvagia S, Donati MA, Funghini S, Ciani F, Pela I, Boneh A, Peters H, Pasquini E, Zammarchi E. Clinical findings and biochemical and molecular analysis of four patients with holocarboxylase synthetase deficiency. *Am J Med Genet*. 2002 Jul 22;111(1):10-8. [\[ABSTRACT\]](#)

Bisanzi S, **Morrone A**, Donati MA, Pasquini E, Spada M, Strisciuglio P, Parenti G, Parini R, Papadia F, Zammarchi E. Genetic analysis in nine unrelated Italian patients affected by OTC deficiency: detection of novel mutations in the OTC gene. *Mol Genet Metab*. 2002 Jun;76(2):137-44. [\[ABSTRACT\]](#)

Funghini S, Pasquini E, Cappellini M, Donati MA, **Morrone A**, Fonda C, Zammarchi E. 3-Hydroxy-3-methylglutaric aciduria in an Italian patient is caused by a new nonsense mutation in the HMGCL gene. *Mol Genet Metab*. 2001 Jul;73(3):268-75. [\[ABSTRACT\]](#)

Porfirio B, Chiarelli I, Graziano C, Mannoni A, **Morrone A**, Zammarchi E, De Bernabé DB, De Córdoba SR. Alkaptonuria in Italy: polymorphic haplotype background, mutational profile, and description of four novel mutations in the homogentisate 1,2-dioxygenase gene. *J Med Genet*. 2000 Apr;37(4):309-12. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

Morrone A, Bardelli T, Donati MA, Giorgi M, Di Rocco M, Gatti R, Parini R, Ricci R, Taddeucci G, D'Azzo A, Zammarchi E. beta-galactosidase gene mutations affecting the lysosomal enzyme and the elastin-binding protein in GM1-gangliosidosis patients with cardiac involvement. Hum Mutat. 2000;15(4):354-66. [\[ABSTRACT\]](#)

Giorgi M, **Morrone A**, Donati MA, Ciani F, Bardelli T, Biasucci G, Zammarchi E. Lymphocyte mRNA analysis of the ornithine transcarbamylase gene in Italian OTCD male patients and manifesting carriers: identification of novel mutations. Hum Mutat. 2000 Apr;15(4):380-1. [\[ABSTRACT\]](#) [\[PDF\]](#)

Morrone A, Pegoraro E, Angelini C, Zammarchi E, Marconi G, Hoffman EP. RNA metabolism in myotonic dystrophy: patient muscle shows decreased insulin receptor RNA and protein consistent with abnormal insulin resistance. J Clin Invest. 1997 Apr 1;99(7):1691-8. [\[ABSTRACT\]](#) [\[FULL TEXT\]](#) [\[PDF\]](#)

Morrone A, Zammarchi E, Scacheri PC, Donati MA, Hoop RC, Servidei S, Galluzzi G, Hoffman EP. Asymptomatic dystrophinopathy. Am J Med Genet. 1997 Mar 31;69(3):261-7. [\[ABSTRACT\]](#)

Zhou XY, van der Spoel A, Rottier R, Hale G, Willemsen R, Berry GT, Strisciuglio P, **Morrone A**, Zammarchi E, Andria G, d'Azzo A. Molecular and biochemical analysis of protective protein/cathepsin A mutations: correlation with clinical severity in galactosialidosis. Hum Mol Genet. 1996 Dec;5(12):1977-87. [\[ABSTRACT\]](#)

Zammarchi E, Donati MA, **Morrone A**, Donzelli GP, Zhou XY, d'Azzo A. Early-infantile galactosialidosis: clinical, biochemical, and molecular observations in a new patient. Am J Med Genet. 1996 Aug 23;64(3):453-8. [\[ABSTRACT\]](#)

Fidzianska A, **Morrone A**, Pegoraro E, Ryniewicz B, Ilnicka A, Zammarchi E, Hoffman EP. An X:autosome translocation stabilizes truncated dystrophin: implications for lack of truncated dystrophins in Duchenne muscular dystrophy. Neuropediatrics. 1995 Jun;26(3):163-7. [\[ABSTRACT\]](#)

Morrone A, Morreau H, Zhou XY, Zammarchi E, Kleijer WJ, Galjaard H, d'Azzo A. Insertion of a T next to the donor splice site of intron 1 causes aberrantly spliced mRNA in a case of infantile GM1-gangliosidosis. Hum Mutat. 1994;3(2):112-20. [\[ABSTRACT\]](#)