

Diebold I, Schön U, Scharf F, et al. Critical assessment of secondary findings in genes linked to primary arrhythmia syndromes. *Human Mutation*. February 2020:humu.23996.

doi:[10.1002/humu.23996](https://doi.org/10.1002/humu.23996)

Wilker M, Christen H-J, Schuster S, Abicht A, Boltshauser E. VLDLR-associated Pontocerebellar Hypoplasia with Nonprogressive Congenital Ataxia and a Diagnostic Neuroimaging Pattern. *Neuropediatrics*. July 2019. doi:[10.1055/s-0039-1688953](https://doi.org/10.1055/s-0039-1688953)

Ikenberg E, Reilich P, Abicht A, Heller C, Schoser B, Walter MC. Charcot-Marie-Tooth disease type 2CC due to a frameshift mutation of the neurofilament heavy polypeptide gene in an Austrian family. *Neuromuscul Disord*. 2019;29(5):392-397. doi:[10.1016/j.nmd.2019.02.007](https://doi.org/10.1016/j.nmd.2019.02.007)

Dusl M, Moreno T, Munell F, et al. Congenital myasthenic syndrome caused by novel COL13A1 mutations. *J Neurol*. 2019;266(5):1107-1112. doi:[10.1007/s00415-019-09239-7](https://doi.org/10.1007/s00415-019-09239-7)

Diebold I, Schön U, Horvath R, et al. HADHA and HADHB gene associated phenotypes - Identification of rare variants in a patient cohort by Next Generation Sequencing. *Mol Cell Probes*. 2019;44:14-20. doi:[10.1016/j.mcp.2019.01.003](https://doi.org/10.1016/j.mcp.2019.01.003)

Kolbel H, Abicht A, Schwartz O, et al. Characteristic clinical and ultrastructural findings in nesprinopathies. *Eur J Paediatr Neurol*. 2019;23(2):254-261. doi:[10.1016/j.ejpn.2018.12.011](https://doi.org/10.1016/j.ejpn.2018.12.011)

Wunderlich G, Abicht A, Brunn A, et al. [Congenital myasthenic syndromes in adulthood : Challenging, rare but treatable]. *Nervenarzt*. 2019;90(2):148-159. doi:[10.1007/s00115-018-0562-9](https://doi.org/10.1007/s00115-018-0562-9)

Thompson R, Abicht A, Beeson D, et al. A nomenclature and classification for the congenital myasthenic syndromes: preparing for FAIR data in the genomic era. *Orphanet J Rare Dis*. 2018;13(1):211. doi:[10.1186/s13023-018-0955-7](https://doi.org/10.1186/s13023-018-0955-7)

Abicht A, Scharf F, Kleinle S, et al. Mitochondrial and nuclear disease panel (Mito-aND-Panel): Combined sequencing of mitochondrial and nuclear DNA by a cost-effective and sensitive NGS-based method. *Mol Genet Genomic Med*. 2018;6(6):1188-1198. doi:[10.1002/mgg3.500](https://doi.org/10.1002/mgg3.500)

Radke J, Dreesmann M, Radke M, et al. The Curse of Apneic Spells. *Semin Pediatr Neurol*. 2018;26:56-58. doi:[10.1016/j.spen.2017.03.006](https://doi.org/10.1016/j.spen.2017.03.006)

Brusa R, Magri F, Papadimitriou D, et al. A new case of limb girdle muscular dystrophy 2G in a Greek patient, founder effect and review of the literature. *Neuromuscul Disord*. 2018;28(6):532-537. doi:[10.1016/j.nmd.2018.04.006](https://doi.org/10.1016/j.nmd.2018.04.006)

Braunisch MC, Gallwitz H, Abicht A, et al. Extension of the phenotype of biallelic loss-of-function mutations in SLC25A46 to the severe form of pontocerebellar hypoplasia type I. *Clin Genet*. 2018;93(2):255-265. doi:[10.1111/cge.13084](https://doi.org/10.1111/cge.13084)

McMacken G, Whittaker RG, Evangelista T, Abicht A, Dusl M, Lochmuller H. Congenital myasthenic syndrome with episodic apnoea: clinical, neurophysiological and genetic features in the long-term follow-up of 19 patients. *J Neurol*. 2018;265(1):194-203.

doi:[10.1007/s00415-017-8689-3](https://doi.org/10.1007/s00415-017-8689-3)

Natera-de Benito D, Topf A, Vilchez JJ, et al. Molecular characterization of congenital myasthenic syndromes in Spain. *Neuromuscul Disord*. 2017;27(12):1087-1098.

doi:[10.1016/j.nmd.2017.08.003](https://doi.org/10.1016/j.nmd.2017.08.003)

Ikenberg E, Karin I, Ertl-Wagner B, et al. Corrigendum to “Rare diagnosis of telethoninopathy (LGMD2G) in a Turkish patient” [Neuromuscular Disorders 27 (2017) 856-860]. *Neuromuscul Disord*. 2017;27(12):e1. doi:[10.1016/j.nmd.2017.10.001](https://doi.org/10.1016/j.nmd.2017.10.001)

Ikenberg E, Karin I, Ertl-Wagner B, et al. Rare diagnosis of telethoninopathy (LGMD2G) in a Turkish patient. *Neuromuscul Disord*. 2017;27(9):856-860. doi:[10.1016/j.nmd.2017.05.017](https://doi.org/10.1016/j.nmd.2017.05.017)

McMacken G, Abicht A, Evangelista T, Spendiff S, Lochmuller H. The Increasing Genetic and Phenotypical Diversity of Congenital Myasthenic Syndromes. *Neuropediatrics*. 2017;48(4):294-308. doi:[10.1055/s-0037-1602832](https://doi.org/10.1055/s-0037-1602832)

Schrank B, Schoser B, Klopstock T, et al. Lifetime exercise intolerance with lactic acidosis as key manifestation of novel compound heterozygous ACAD9 mutations causing complex I deficiency. *Neuromuscul Disord*. 2017;27(5):473-476. doi:[10.1016/j.nmd.2017.02.005](https://doi.org/10.1016/j.nmd.2017.02.005)

Elbracht M, Mull M, Wagner N, et al. Stroke as Initial Manifestation of Adenosine Deaminase 2 Deficiency. *Neuropediatrics*. 2017;48(2):111-114. doi:[10.1055/s-0036-1597611](https://doi.org/10.1055/s-0036-1597611)

Luhl S, Bode H, Schlotzer W, et al. Novel homozygous RARS2 mutation in two siblings without pontocerebellar hypoplasia - further expansion of the phenotypic spectrum. *Orphanet J Rare Dis*. 2016;11(1):140. doi:[10.1186/s13023-016-0525-9](https://doi.org/10.1186/s13023-016-0525-9)

O'Connor E, Topf A, Muller JS, et al. Identification of mutations in the MYO9A gene in patients with congenital myasthenic syndrome. *Brain*. 2016;139(Pt 8):2143-2153. doi:[10.1093/brain/aww130](https://doi.org/10.1093/brain/aww130)

Claeys KG, Abicht A, Hausler M, et al. Novel genetic and neuropathological insights in neurogenic muscle weakness, ataxia, and retinitis pigmentosa (NARP). *Muscle Nerve*. 2016;54(2):328-333. doi:[10.1002/mus.25125](https://doi.org/10.1002/mus.25125)

Lane M, Boczonadi V, Bachtari S, et al. Mitochondrial dysfunction in liver failure requiring transplantation. *J Inherit Metab Dis*. 2016;39(3):427-436. doi:[10.1007/s10545-016-9927-z](https://doi.org/10.1007/s10545-016-9927-z)

Derevenciuc A-I, Abicht A, Hamza S, Roth C, Ferbert A. Thomsen myotonia--A 4-generation family with a new mutation and a mild phenotype. *Muscle Nerve*. 2016;53(4):653-654. doi:[10.1002/mus.24971](https://doi.org/10.1002/mus.24971)

Suriyanarayanan S, Auranen M, Toppila J, et al. The Variant p.(Arg183Trp) in SPTLC2 Causes Late-Onset Hereditary Sensory Neuropathy. *Neuromolecular Med.* 2016;18(1):81-90. doi:[10.1007/s12017-015-8379-1](https://doi.org/10.1007/s12017-015-8379-1)

Natera-de Benito D, Nascimento A, Abicht A, et al. KLHL40-related nemaline myopathy with a sustained, positive response to treatment with acetylcholinesterase inhibitors. *J Neurol.* 2016;263(3):517-523. doi:[10.1007/s00415-015-8015-x](https://doi.org/10.1007/s00415-015-8015-x)

Natera-de Benito D, Bestue M, Vilchez JJ, et al. Long-term follow-up in patients with congenital myasthenic syndrome due to RAPSN mutations. *Neuromuscul Disord.* 2016;26(2):153-159. doi:[10.1016/j.nmd.2015.10.013](https://doi.org/10.1016/j.nmd.2015.10.013)

Kohler C, Heyer C, Hoffjan S, et al. Early-onset leukoencephalopathy due to a homozygous missense mutation in the DARS2 gene. *Mol Cell Probes.* 2015;29(5):319-322. doi:[10.1016/j.mcp.2015.06.005](https://doi.org/10.1016/j.mcp.2015.06.005)

Dusl M, Senderek J, Muller JS, et al. A 3'-UTR mutation creates a microRNA target site in the GFPT1 gene of patients with congenital myasthenic syndrome. *Hum Mol Genet.* 2015;24(12):3418-3426. doi:[10.1093/hmg/ddv090](https://doi.org/10.1093/hmg/ddv090)

Orstavik K, Wallace SC, Torbergesen T, et al. A de novo Mutation in the SCN4A Gene Causing Sodium Channel Myotonia. *J Neuromuscul Dis.* 2015;2(2):181-184. doi:[10.3233/JND-150069](https://doi.org/10.3233/JND-150069)

Pyle A, Nightingale HJ, Griffin H, et al. Respiratory chain deficiency in nonmitochondrial disease. *Neurol Genet.* 2015;1(1):e6. doi:[10.1212/NXG.0000000000000006](https://doi.org/10.1212/NXG.0000000000000006)

Braczynski AK, Vlaho S, Muller K, et al. ATP synthase deficiency due to TMEM70 mutation leads to ultrastructural mitochondrial degeneration and is amenable to treatment. *Biomed Res Int.* 2015;2015:462592. doi:[10.1155/2015/462592](https://doi.org/10.1155/2015/462592)

Nicole S, Chaouch A, Torbergesen T, et al. Agrin mutations lead to a congenital myasthenic syndrome with distal muscle weakness and atrophy. *Brain.* 2014;137(Pt 9):2429-2443. doi:[10.1093/brain/awu160](https://doi.org/10.1093/brain/awu160)

Taylor RW, Pyle A, Griffin H, et al. Use of whole-exome sequencing to determine the genetic basis of multiple mitochondrial respiratory chain complex deficiencies. *JAMA.* 2014;312(1):68-77. doi:[10.1001/jama.2014.7184](https://doi.org/10.1001/jama.2014.7184)

Rosenbohm A, Sussmuth SD, Kassubek J, et al. Novel ETFDH mutation and imaging findings in an adult with glutaric aciduria type II. *Muscle Nerve.* 2014;49(3):446-450. doi:[10.1002/mus.23979](https://doi.org/10.1002/mus.23979)

Dilena R, Abicht A, Sergi P, et al. Congenital myasthenic syndrome due to choline acetyltransferase mutations in infants: clinical suspicion and comprehensive electrophysiological assessment are important for early diagnosis. *J Child Neurol.* 2014;29(3):389-393. doi:[10.1177/0883073812470000](https://doi.org/10.1177/0883073812470000)

Schreckenbach T, Schroder JM, Voit T, et al. Novel TPM3 mutation in a family with cap myopathy and review of the literature. *Neuromuscul Disord.* 2014;24(2):117-124. doi:[10.1016/j.nmd.2013.10.002](https://doi.org/10.1016/j.nmd.2013.10.002)

Gallenmuller C, Muller-Felber W, Dusl M, et al. Salbutamol-responsive limb-girdle congenital myasthenic syndrome due to a novel missense mutation and heteroallelic deletion in MUSK. *Neuromuscul Disord.* 2014;24(1):31-35. doi:[10.1016/j.nmd.2013.08.002](https://doi.org/10.1016/j.nmd.2013.08.002)

Chaouch A, Porcelli V, Cox D, et al. Mutations in the Mitochondrial Citrate Carrier SLC25A1 are Associated with Impaired Neuromuscular Transmission. *J Neuromuscul Dis.* 2014;1(1):75-90. doi:[10.3233/JND-140021](https://doi.org/10.3233/JND-140021)

Kipfer S, Jung S, Lemke JR, et al. Novel CACNA1A mutation(s) associated with slow saccade velocities. *J Neurol.* 2013;260(12):3010-3014. doi:[10.1007/s00415-013-7099-4](https://doi.org/10.1007/s00415-013-7099-4)

Peyer A-K, Abicht A, Heinimann K, Sinnreich M, Fischer D. Quinine sulfate as a therapeutic option in a patient with slow channel congenital myasthenic syndrome. *Neuromuscul Disord.* 2013;23(7):571-574. doi:[10.1016/j.nmd.2013.04.001](https://doi.org/10.1016/j.nmd.2013.04.001)

Mahjneh I, Lochmuller H, Muntoni F, Abicht A. DOK7 limb-girdle myasthenic syndrome mimicking congenital muscular dystrophy. *Neuromuscul Disord.* 2013;23(1):36-42. doi:[10.1016/j.nmd.2012.06.355](https://doi.org/10.1016/j.nmd.2012.06.355)

Marina AD, Schara U, Pyle A, et al. NDUFS8-related Complex I Deficiency Extends Phenotype from “PEO Plus” to Leigh Syndrome. *JIMD Rep.* 2013;10:17-22. doi:[10.1007/8904_2012_195](https://doi.org/10.1007/8904_2012_195)

Czell D, Abicht A, Hench J, Weber M. Exercise-induced myalgia and rhabdomyolysis in a patient with the rare m.3243A>T mtDNA mutation. *BMJ Case Rep.* 2012;2012. doi:[10.1136/bcr-2012-006980](https://doi.org/10.1136/bcr-2012-006980)

Neeve VCM, Samuels DC, Bindoff LA, et al. What is influencing the phenotype of the common homozygous polymerase-gamma mutation p.Ala467Thr? *Brain.* 2012;135(Pt 12):3614-3626. doi:[10.1093/brain/aws298](https://doi.org/10.1093/brain/aws298)

Abicht A, Dusl M, Gallenmuller C, et al. Congenital myasthenic syndromes: achievements and limitations of phenotype-guided gene-after-gene sequencing in diagnostic practice: a study of 680 patients. *Hum Mutat.* 2012;33(10):1474-1484. doi:[10.1002/humu.22130](https://doi.org/10.1002/humu.22130)

Bulst S, Holinski-Feder E, Payne B, et al. In vitro supplementation with deoxynucleoside monophosphates rescues mitochondrial DNA depletion. *Mol Genet Metab.* 2012;107(1-2):95-103. doi:[10.1016/j.ymgme.2012.04.022](https://doi.org/10.1016/j.ymgme.2012.04.022)

van Baalen A, Hausler M, Plecko-Startinig B, et al. Febrile infection-related epilepsy syndrome without detectable autoantibodies and response to immunotherapy: a case series and discussion of epileptogenesis in FIRES. *Neuropediatrics.* 2012;43(4):209-216. doi:[10.1055/s-0032-1323848](https://doi.org/10.1055/s-0032-1323848)

Schara U, Della Marina A, Abicht A. Congenital myasthenic syndromes: current diagnostic and therapeutic approaches. *Neuropediatrics.* 2012;43(4):184-193. doi:[10.1055/s-0032-1323850](https://doi.org/10.1055/s-0032-1323850)

Guergueltecheva V, Muller JS, Dusl M, et al. Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. *J Neurol.* 2012;259(5):838-850.

doi:[10.1007/s00415-011-6262-z](https://doi.org/10.1007/s00415-011-6262-z)

Brackmann F, Abicht A, Ahting U, Schroder R, Trollmann R. Classical MERRF phenotype associated with mitochondrial tRNA(Leu) (m.3243A>G) mutation. *Eur J Pediatr.*

2012;171(5):859-862. doi:[10.1007/s00431-011-1662-8](https://doi.org/10.1007/s00431-011-1662-8)

Chaouch A, Muller JS, Guergueltecheva V, et al. A retrospective clinical study of the treatment of slow-channel congenital myasthenic syndrome. *J Neurol.* 2012;259(3):474-481.

doi:[10.1007/s00415-011-6204-9](https://doi.org/10.1007/s00415-011-6204-9)

Abicht A, Kroger S, Schoser B. [Neuromuscular signal transmission in adulthood. Current facets of acquired and hereditary disorders]. *Nervenarzt.* 2011;82(6):707-711.

doi:[10.1007/s00115-010-2969-9](https://doi.org/10.1007/s00115-010-2969-9)

Senderek J, Muller JS, Dusl M, et al. Hexosamine biosynthetic pathway mutations cause neuromuscular transmission defect. *Am J Hum Genet.* 2011;88(2):162-172.

doi:[10.1016/j.ajhg.2011.01.008](https://doi.org/10.1016/j.ajhg.2011.01.008)

Schara U, von Kleist-Retzow J-C, Lainka E, et al. Acute liver failure with subsequent cirrhosis as the primary manifestation of TRMU mutations. *J Inherit Metab Dis.* 2011;34(1):197-201.

doi:[10.1007/s10545-010-9250-z](https://doi.org/10.1007/s10545-010-9250-z)

Kemp JP, Smith PM, Pyle A, et al. Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency. *Brain.* 2011;134(Pt 1):183-195.

doi:[10.1093/brain/awq320](https://doi.org/10.1093/brain/awq320)

Seeger J, Schrank B, Pyle A, et al. Clinical and neuropathological findings in patients with TACO1 mutations. *Neuromuscul Disord.* 2010;20(11):720-724. doi:[10.1016/j.nmd.2010.06.017](https://doi.org/10.1016/j.nmd.2010.06.017)

Walter MC, Czermin B, Muller-Ziermann S, et al. Late-onset ptosis and myopathy in a patient with a heterozygous insertion in POLG2. *J Neurol.* 2010;257(9):1517-1523.

doi:[10.1007/s00415-010-5565-9](https://doi.org/10.1007/s00415-010-5565-9)

Mihaylova V, Scola RH, Gervini B, et al. Molecular characterisation of congenital myasthenic syndromes in Southern Brazil. *J Neurol Neurosurg Psychiatry.* 2010;81(9):973-977.

doi:[10.1136/jnnp.2009.177816](https://doi.org/10.1136/jnnp.2009.177816)

Schara U, Christen H-J, Durmus H, et al. Long-term follow-up in patients with congenital myasthenic syndrome due to CHAT mutations. *Eur J Paediatr Neurol.* 2010;14(4):326-333.

doi:[10.1016/j.ejpn.2009.09.009](https://doi.org/10.1016/j.ejpn.2009.09.009)

Martin P, Rautenstraubeta B, Abicht A, Fahrbach J, Koster S. Severe Myoclonic Epilepsy in Infancy - Adult Phenotype with Bradykinesia, Hypomimia, and Perseverative Behavior: Report of Five Cases. *Mol Syndromol.* 2010;1(5):231-238. doi:[10.1159/000326746](https://doi.org/10.1159/000326746)

Mihaylova V, Salih MAM, Mukhtar MM, et al. Refinement of the clinical phenotype in musk-related congenital myasthenic syndromes. *Neurology*. 2009;73(22):1926-1928.

doi:[10.1212/WNL.0b013e3181c3fce9](https://doi.org/10.1212/WNL.0b013e3181c3fce9)

Schara U, Barisic N, Deschauer M, et al. Ephedrine therapy in eight patients with congenital myasthenic syndrome due to DOK7 mutations. *Neuromuscul Disord*. 2009;19(12):828-832.

doi:[10.1016/j.nmd.2009.09.008](https://doi.org/10.1016/j.nmd.2009.09.008)

Bulst S, Abicht A, Holinski-Feder E, et al. In vitro supplementation with dAMP/dGMP leads to partial restoration of mtDNA levels in mitochondrial depletion syndromes. *Hum Mol Genet*. 2009;18(9):1590-1599. doi:[10.1093/hmg/ddp074](https://doi.org/10.1093/hmg/ddp074)

Horvath R, Bender A, Abicht A, et al. Heteroplasmic mutation in the anticodon-stem of mitochondrial tRNA(Val) causing. *J Neurol*. 2009;256(5):810-815. doi:[10.1007/s00415-009-5023-8](https://doi.org/10.1007/s00415-009-5023-8)

Feddersen B, DE LA Fontaine L, Sass JO, et al. Mitochondrial neurogastrointestinal encephalomyopathy mimicking anorexia nervosa. *Am J Psychiatry*. 2009;166(4):494-495.

doi:[10.1176/appi.ajp.2008.08101525](https://doi.org/10.1176/appi.ajp.2008.08101525)

Mihaylova V, Muller JS, Vilchez JJ, et al. Clinical and molecular genetic findings in COLQ-mutant congenital myasthenic syndromes. *Brain*. 2008;131(Pt 3):747-759.

doi:[10.1093/brain/awm325](https://doi.org/10.1093/brain/awm325)

Muller JS, Mihaylova V, Abicht A, Lochmuller H. Congenital myasthenic syndromes: spotlight on genetic defects of neuromuscular transmission. *Expert Rev Mol Med*. 2007;9(22):1-20.

doi:[10.1017/S1462399407000427](https://doi.org/10.1017/S1462399407000427)

Muller JS, Herczegfalvi A, Vilchez JJ, et al. Phenotypical spectrum of DOK7 mutations in congenital myasthenic syndromes. *Brain*. 2007;130(Pt 6):1497-1506. doi:[10.1093/brain/awm068](https://doi.org/10.1093/brain/awm068)

Muller JS, Baumeister SK, Rasic VM, et al. Impaired receptor clustering in congenital myasthenic syndrome with novel RAPSN mutations. *Neurology*. 2006;67(7):1159-1164.

doi:[10.1212/01.wnl.0000233837.79459.40](https://doi.org/10.1212/01.wnl.0000233837.79459.40)

Muller JS, Baumeister SK, Schara U, et al. CHRND mutation causes a congenital myasthenic syndrome by impairing co-clustering of the acetylcholine receptor with rapsyn. *Brain*. 2006;129(Pt 10):2784-2793. doi:[10.1093/brain/awl188](https://doi.org/10.1093/brain/awl188)

Colomer J, Muller JS, Vernet A, et al. Long-term improvement of slow-channel congenital myasthenic syndrome with fluoxetine. *Neuromuscul Disord*. 2006;16(5):329-333.

doi:[10.1016/j.nmd.2006.02.009](https://doi.org/10.1016/j.nmd.2006.02.009)

von der Hagen M, Schallner J, Kaindl AM, et al. Facing the genetic heterogeneity in neuromuscular disorders: linkage analysis as an economic diagnostic approach towards the molecular diagnosis. *Neuromuscul Disord*. 2006;16(1):4-13. doi:[10.1016/j.nmd.2005.10.001](https://doi.org/10.1016/j.nmd.2005.10.001)

Horvath R, Abicht A, Holinski-Feder E, et al. Leigh syndrome caused by mutations in the flavoprotein (Fp) subunit of succinate dehydrogenase (SDHA). *J Neurol Neurosurg Psychiatry*. 2006;77(1):74-76. doi:[10.1136/jnnp.2005.067041](https://doi.org/10.1136/jnnp.2005.067041)

Muller JS, Stucka R, Neudecker S, et al. An intronic base alteration of the CHRNE gene leading to a congenital myasthenic syndrome. *Neurology*. 2005;65(3):463-465. doi:[10.1212/01.wnl.0000172346.26219.fd](https://doi.org/10.1212/01.wnl.0000172346.26219.fd)

Soltanzadeh P, Muller JS, Ghorbani A, Abicht A, Lochmuller H, Soltanzadeh A. An Iranian family with congenital myasthenic syndrome caused by a novel acetylcholine receptor mutation (CHRNE K171X). *J Neurol Neurosurg Psychiatry*. 2005;76(7):1039-1040. doi:[10.1136/jnnp.2004.059436](https://doi.org/10.1136/jnnp.2004.059436)

Barisic N, Muller JS, Paucic-Kirincic E, et al. Clinical variability of CMS-EA (congenital myasthenic syndrome with episodic apnea) due to identical CHAT mutations in two infants. *Eur J Paediatr Neurol*. 2005;9(1):7-12. doi:[10.1016/j.ejpn.2004.10.008](https://doi.org/10.1016/j.ejpn.2004.10.008)

Muller JS, Abicht A, Christen H-J, et al. A newly identified chromosomal microdeletion of the rapsyn gene causes a congenital myasthenic syndrome. *Neuromuscul Disord*. 2004;14(11):744-749. doi:[10.1016/j.nmd.2004.06.010](https://doi.org/10.1016/j.nmd.2004.06.010)

Morar B, Gresham D, Angelicheva D, et al. Mutation history of the roma/gypsies. *Am J Hum Genet*. 2004;75(4):596-609. doi:[10.1086/424759](https://doi.org/10.1086/424759)

Veitenhansl M, Stegner K, Hierl F-X, et al. 40(th) EASD Annual Meeting of the European Association for the Study of Diabetes : Munich, Germany, 5-9 September 2004. *Diabetologia*. 2004;47(Suppl 1):A1-A464. doi:[10.1007/BF03375463](https://doi.org/10.1007/BF03375463)

Muller JS, Abicht A, Burke G, et al. The congenital myasthenic syndrome mutation RAPSN N88K derives from an ancient Indo-European founder. *J Med Genet*. 2004;41(8):e104. doi:[10.1136/jmg.2004.021139](https://doi.org/10.1136/jmg.2004.021139)

Muller JS, Petrova S, Kiefer R, et al. Synaptic congenital myasthenic syndrome in three patients due to a novel missense mutation (T441A) of the COLQ gene. *Neuropediatrics*. 2004;35(3):183-189. doi:[10.1055/s-2004-820996](https://doi.org/10.1055/s-2004-820996)

Muller JS, Mildner G, Muller-Felber W, et al. Rapsyn N88K is a frequent cause of congenital myasthenic syndromes in European patients. *Neurology*. 2003;60(11):1805-1810. doi:[10.1212/01.wnl.0000072262.14931.80](https://doi.org/10.1212/01.wnl.0000072262.14931.80)

Schmidt C, Abicht A, Krampfl K, et al. Congenital myasthenic syndrome due to a novel missense mutation in the gene encoding choline acetyltransferase. *Neuromuscul Disord*. 2003;13(3):245-251.

Abicht A, Lochmuller H. What's in the serum of seronegative MG and LEMS?: MuSK et al. *Neurology*. 2002;59(11):1672-1673. doi:[10.1212/01.wnl.0000041026.90947.79](https://doi.org/10.1212/01.wnl.0000041026.90947.79)

Barisic N, Schmidt C, Sidorova OP, et al. Congenital myasthenic syndrome (CMS) in three European kinships due to a novel splice mutation (IVS7 - 2 A/G) in the epsilon acetylcholine receptor (AChR) subunit gene. *Neuropediatrics*. 2002;33(5):249-254. doi:[10.1055/s-2002-36738](https://doi.org/10.1055/s-2002-36738)

Abicht A, Stucka R, Schmidt C, et al. A newly identified chromosomal microdeletion and an N-box mutation of the AChR epsilon gene cause a congenital myasthenic syndrome. *Brain*. 2002;125(Pt 5):1005-1013. doi:[10.1093/brain/awf095](https://doi.org/10.1093/brain/awf095)

Jahn K, Mohammadi B, Krampfl K, Abicht A, Lochmuller H, Bufler J. Deactivation and desensitization of mouse embryonic- and adult-type nicotinic receptor channel currents. *Neurosci Lett*. 2001;307(2):89-92. doi:[10.1016/s0304-3940\(01\)01929-2](https://doi.org/10.1016/s0304-3940(01)01929-2)

Abicht A, Lochmuller H. Technology evaluation: edrecolomab, Centocor Inc. *Curr Opin Mol Ther*. 2000;2(5):593-600.

Stucka R, Abicht A, Song IH, Bonsch D, Deufel T, Lochmuller H. A modified alignment of human and rodent 5' untranslated sequences of the acetylcholine receptor epsilon subunit gene reveals additional regions of high homology. *Neuromuscul Disord*. 2000;10(3):213-214.

Horvath R, Abicht A, Shoubridge EA, et al. Leber's hereditary optic neuropathy presenting as multiple sclerosis-like disease of the CNS. *J Neurol*. 2000;247(1):65-67.

Abicht A, Stucka R, Karcagi V, et al. A common mutation (epsilon1267delG) in congenital myasthenic patients of Gypsy ethnic origin. *Neurology*. 1999;53(7):1564-1569. doi:[10.1212/wnl.53.7.1564](https://doi.org/10.1212/wnl.53.7.1564)

Abicht A, Lochmuller H. Technology evaluation: CRIB (CNTF delivery) CytoTherapeutics Inc. *Curr Opin Mol Ther*. 1999;1(5):645-650.

Toepfer M, Fischer P, Abicht A, Lochmuller H, Pongratz D, Muller-Felber W. Localization of transforming growth factor beta in association with neuromuscular junctions in adult human muscle. *Cell Mol Neurobiol*. 1999;19(2):297-300.

Kramer R, Lochmuller H, Abicht A, Rudel R, Brinkmeier H. Myotonic ADR-MDX mutant mice show less severe muscular dystrophy than MDX mice. *Neuromuscul Disord*. 1998;8(8):542-550.

Abicht A, Muller-Felber W, Fischer P, et al. Congenital myasthenic syndromes: clinical and genetic analysis of 18 patients. *Eur J Med Res*. 1997;2(12):515-522.

Abicht A, Muller JJ, Lochmuller H. Congenital Myasthenic Syndromes. In: Adam MP, Ardinger HH, Pagon RA, et al., eds. *GeneReviews((R))*. Seattle (WA): University of Washington, Seattle; 1993.