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| --- | --- | --- | --- | --- | --- | --- |
|  | **Accession Number** | **Codon number** | **Codon change** | **Amino acid change** | **CDNA change: NM\_000158:** | **Reference** |
| Point mutations (missense and nonsense) |
| 1 | CM0910742 | 152 | GAG-TAG | p.E152X | c.454G>T | Lamperti (2009) J Inherit Metab Dis **32S1,** S161 Additional report available to subscribers |
| 2 | CM100317[rs773558446](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=773558446) | 188 | CAT-CCT | p.H188P | c.563A>C | [Fernandez (2010) Muscle Nerve 41, 269](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=19813197&dopt=Abstract) |
| 3 | CM960703[rs137852886](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=137852886) | 224 | CTA-CCA | p.L224P | c.671T>C | Bao (1996) J Clin Invest **97,** 941  |
| 4 | CM061009[rs137852892](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=137852892) | 236 | CAG-CAC | p.Q236H | c.708G>C | [Burrow (2006) Am J Med Genet A 140A, 878](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=16528737&dopt=Abstract) |
| 5 | CM042990 | 243 | CAT-CGT | p.H243R | c.728A>G | [Bruno (2004) Neurology 63, 1053](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=15452297&dopt=Abstract) |
| 6 | CM960704[rs137852887](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=137852887) | 257 | TTT-TTA | p.F257L | c.771T>A | [Bao (1996) J Clin Invest 97, 941](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=8613547&dopt=Abstract) |
| 7 | CM061010[rs369574719](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=369574719) R to H [rs137852893](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=137852893) | 262 | CGT-TGT | p.R262C | c.784C>T | [Burrow (2006) Am J Med Genet A 140A, 878](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=16528737&dopt=Abstract) |
| 8 | CM053896[rs779475367](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=779475367) | 299 | GGA-TGA | p.G299X | c.895G>T | L'hermine-Coulomb (2005) Am J Med Genet A **139A,** 118  |
| 9 | CM960705[rs80338671](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=80338671) | 329 | TAC-TCC | p.Y329S | c.986A>C | Bao (1996) J Clin Invest **97,** 941  |
| 10 | CM121171 | 427 | GGA-AGA | p.G427R | c.1279G>A | [Magoulas (2012) Hum Pathol epub, epub](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=22305237&dopt=Abstract) |
| 11 | CM106724[rs1456579860](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=1456579860) | 495 | ATG-ACG | p.M495T | c.1484T>C | [Li (2010) J Inherit Metab Dis ,](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=20058079&dopt=Abstract)  |
| 12 | CM000705[rs201958741](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=201958741) | 515 | CGT-CAT | p.R515H | c.1544G>A | Ziemssen (2000) Ann Neurol **47,** 536  |
| 13 | CM960706[rs80338672](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=80338672) | 515 | CGT-TGT | p.R515C | c.1543C>T | [Bao (1996) J Clin Invest 97, 941](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=8613547&dopt=Abstract) |
| 14 | CM993166[rs80338673](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=80338673) | 524 | CGA-CAA | p.R524Q | c.1571G>A | Bruno (1999) Neuromuscul Disord **9,** 403  |
| 15 | CM960707[rs137852888](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=137852888) | 524 | CGA-TGA | p.R524X | c.1570C>T | Bao (1996) J Clin Invest **97,** 941  |
| 16 | CM081269[rs886058900](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=886058900) | 535 | TAT-TGT | p.Y535C | c.1604A>G | Massa (2008) Muscle Nerve **37,** 530 Additional report available to subscribers |
| 17 | CM042991[rs137852889](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=137852889) | 545 | CAT-CGT | p.H545R | c.1634A>G | [Bruno (2004) Neurology 63, 1053](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=15452297&dopt=Abstract) |
| 18 | CM073083[rs137852894](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=137852894) | 548 | TGG-TAG | p.W548X | c.1643G>A | Assereto (2007) Biochem Biophys Res Commun **361,** 445 Additional report available to subscribers |
| 19 | CM106725[rs777589783](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=777589783) | 552 | CCA-CTA | p.P552L | c.1655C>T | [Li (2010) J Inherit Metab Dis ,](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=20058079&dopt=Abstract)  |
| 20 | CM042992[rs137852890](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=137852890) | 592 | GAA-TAA | p.E592X | c.1774G>T | Bruno (2004) Neurology **63,** 1053 Additional report available to subscribers |
| 21 | CM042993[rs137852891](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=137852891) | 628 | CAT-CGT | p.H628R | c.1883A>G | [Bruno (2004) Neurology 63, 1053](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=15452297&dopt=Abstract) |
| 22 | CM042994[rs766935302](https://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=766935302) | 637 | CGA-TGA | p.R637X | c.1909C>T | Bruno (2004) Neurology **63,** 1053 Additional report available to subscribers |

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| Splice site mutations |
| 23 | CS043130 | IVS1 ds +1 G>A | [Bruno (2004) Neurology 63, 1053](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=15452297&dopt=Abstract) |
| 24 | CS100318 | IVS5 ds +2 T>C | [Fernandez (2010) Muscle Nerve 41, 269](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=19813197&dopt=Abstract) |
| 25 | CS073485 | IVS5 ds +5 G>C | Assereto (2007) Biochem Biophys Res Commun 361, 445 |
| 26 | CS961594 | IVS6 as -1 G>A | Bao (1996) J Clin Invest 97, 941 |
| 27 | CS108956 | IVS9 ds +1 G>C | Rothacker (2010) Pathologe 31, 293 |
| 28 | CS067093 | IVS10 as -1 G>A | [Akman (2006) Prenat Diagn 26, 951](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=16874838&dopt=Abstract) |
| Gross Deletions |
| 29 | CG082366 | ex. 16 (described at genomic DNA level) | [Raju (2008) J Child Neurol **23,** 349](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=18230843&dopt=Abstract) |
| 30 | CG044348 | ex. 4-6 (described at genomic DNA level) | [Bruno (2004) Neurology **63,** 1053](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=15452297&dopt=Abstract) |
| 31 | CG044349 | ex. 8-12 (described at genomic DNA level) | [Tay (2004) Neuromuscul Disord **14,** 253](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=15019703&dopt=Abstract) |
| 32 | CG123342 | incl. ex. 2-7 (described at genomic DNA level) | [Li (2012) J Child Neurol **27,** 204](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=21917543&dopt=Abstract) |
| 33 | CG123341 | incl. ex. 7 (described at genomic DNA level) | [Li (2012) J Child Neurol **27,** 204](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=21917543&dopt=Abstract) |
| Small Deletions |
| 34 | CD035910 | CGGGA^(95)GCAGAaGGAGTTTTTC | [Nambu (2003) Neurology **61,** 392](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=12913206&dopt=Abstract) |
| 35 | CD040898 | TGTTGTAG\_T9E10\_GAt^(414)GTATCAGGAA | [Tay (2004) Neuromuscul Disord **14,** 253](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=15019703&dopt=Abstract) |
| 36 | CD040899 | GGATAAG^(489)TCGcTGGCATTTTG | [Tay (2004) Neuromuscul Disord **14,** 253](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=15019703&dopt=Abstract) |
| 37 | CD106726 | GGACCAC^(666)AGCaCTGACTTTTT | [Li (2010) J Inherit Metab Dis **,**](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=20058079&dopt=Abstract)  |
| Complex rearrangements  |
| 38 | CP107909 | Del 28bp, ins G in ex. 5 | [Taratuto (2010) Neuromuscul Disord **20,** 783](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=20833045&dopt=Abstract) |
| 39 | CP086734 | g.101517\_127067del25550insCAGTACTAA, del ex. 4-7 | [Nolte (2008) Acta Neuropathol **116,** 491](http://www.ncbi.nlm.nih.gov/sites/entrez?cmd=Retrieve&db=PubMed&list_uids=18661138&dopt=Abstract) |
| Deep Intronic mutations |
| 40 | [rs869320698](https://www.ncbi.nlm.nih.gov/variation/tools/1000genomes/?chr=3&from=81542964&to=81542972&gts=rs869320698&mk=81542964:81542972|rs869320698) | c.2052+5289\_5297delinsTGTTTTTTACATGACAGGT | [Akman et al (2015) JAMA Neurology Network](http://archneur.jamanetwork.com/article.aspx?articleid=2099526)  |