

Exon	Variant	Reference
1	<b>p.L13R</b>	<b>This study</b>
2	<b>p.D17del</b>	<b>This study</b>
	p.C35R p.R44C	Lynch et al. Anesthesiology 1997; 86:620-6 Tammaro et al. Clin Chem 2003; 49:761-8
3	<b>p.D60N</b>	<b>Wu et al. Brain 2006 (in press).</b>
	p.S71Y	Zhou et al. Neuromuscul Disord 2005; 15:676–743
4	p.R109W	Zhou et al. Neuromuscul Disord 2005; 15:676–743
6	<b>p.Q155K</b>	<b>This study</b>
	p.E160G	Shepherd et al. J Med Genet 2004; 41:e33
	<b>p.R163C</b>	Quane et al. Nat Genet 1993; 5:51-5
	p.R163L	Monnier et al. Hum Mutat 2005; 26:413-25
	p.G165R	Monnier et al. Hum Mutat 2005; 26:413-25
	<b>p.D166G</b>	<b>This study</b>
6	<b>p.D166N</b>	Rueffert et al. Acta Anaesthesiol Scand 2002; 46:692-8
	p.R177C	Monnier et al. Hum Mutat 2005; 26:413-25
	p.Y178C	Monnier et al. Hum Mutat 2005; 26:413-25
7	p.G215E	Zhou et al. Neuromuscul Disord 2005; 15:676–743
8	<b>p.V218I</b>	<b>This study</b>
	p.D227V	Monnier et al. Hum Mutat 2005; 26:413-25
9	p.G248R	Gillard et al. Genomics 1992; 13:1247-54
10	<b>p.R316L</b>	<b>This study</b>
11	p.R328W	Loke et al. Anesthesiology 2003; 99:297-302
	<b>p.G341R</b>	Quane et al. Hum Mol Genet 1994; 3:471-6
12	p.R401C	Davis et al. Br J Anaesth 2002; 88:508-15

Exon	Variant	Reference
12	p.R401H	Rueffert et al. Acta Anaesthesiol Scand 2002; 46:692-8
	p.R401S	Monnier et al. Hum Mutat 2005; 26:413-25
	p.M402I	Zhou et al. Neuromuscul Disord 2005; 15:676–743
	p.I403M	Quane et al. Nat Genet 1993; 5:51-5
13	<b>p.S427L</b>	<b>Wu et al. Brain 2006 (in press).</b>
	p.R471C	Gillard et al. Genomics 1992; 13:1247-54
14	<b>p.Q474H</b>	<b>This study</b>
	<b>p.E512K</b>	<b>Wu et al. Brain 2006 (in press).</b>
	p.Y522S p.Y522C	Quane et al. Genomics 1994; 23:236-9 Yeh et al. Anesth Analg 2005; 101:1401-6
15	<b>p.R533H</b>	Brandt et al. Hum Mol Genet 1999; 8:2055-62
	p.R533C p.R552W	Tammaro et al. Clin Chem 2003; 49:761-8 Keating et al. J Med Genet 1997; 34:291-6
17	p.R614L	Quane et al. Br J Anaesth 1997; 79:332-7
	p.R614C	Gillard et al. Genomics 1991; 11:751-5
19	p.N759D	Kossugue et al. Neuromuscul.Disord 2005; 15:676–743
33	<b>p.P1592L</b>	<b>This study</b>
	R1606C	Jungbluth et al. Neuromuscul Disord 2005; 15:676–743.
34	<b>p.R1667C</b>	<b>This study</b>
	p.S1728F	Sambuughin et al. Anesthesiology 2005; 102:515-21
	<b>p.P1763S</b>	<b>This study</b>
	<b>p.P1773S</b>	<b>This study</b>
	p.L1786P	Gillard et al. Genomics 1992; 13:1247-54
	p.P1787L	Gillard et al. Genomics 1992; 13:1247-54

Exon	Variant	Reference
38	p.G2060C	Gillard et al. Genomics 1992; 13:1247-54
39	p.M2101L	Tammaro et al. Clin Chem 2003; 49:761-8
	p.V2117L	Tammaro et al. Clin Chem 2003; 49:761-8
	p.D2129E	Rueffert et al. Hum Mutat 2001; 17:238
	p.R2163P	Fortunato et al. Eur J Hum Genet 2000 ; 8:149-52
	p.R2163C	Manning et al. Am J Hum Genet 1998; 62:599-609
	<b>p.R2163H</b>	Manning et al. Am J Hum Genet 1998; 62:599-609
40	p.V2168M	Manning et al. Am J Hum Genet 1998; 62:599-609
	p.I2182F	Ruffert et al. Anaesthesist 2002; 51:904-13
	p.A2200V	Sambuughin et al. Anesthesiology 2005; 102:515-21
	p.T2206M	Manning et al. Am J Hum Genet 1998; 62:599-609
	p.T2206R	Brandt et al. Hum Mol Genet 1999; 8:2055-62
42	p.V2210F	Sambuughin et al. Anesthesiology 2005; 102:515-21
	p.V2214I	Sambuughin et al. Anesthesiology. 2001; 95:594-9
42	p.V2280I	Galli et al. Cell Calcium 2002; 32:143-51
	p.N2283H	Zhou et al. Neuromuscul Disord 2005; 15:676–743
43	p.N2342S	Marchant et al. Muscle Nerve 2004; 30:114-7
44	p.E2344D	Monnier et al. Hum Mutat 2005; 26:413-25
	p.V2346M	Shepherd et al. J Med Genet 2004; 41:e33
	p.E2347del	Sambuughin et al. Am J Hum Genet 2001; 69:204-8
	p.E2348G	Shepherd et al. J Med Genet 2004; 41:e33
	p.A2350T	Sambuughin et al. Neuromuscul Disord 2001; 11:530-7
	p.R2355C	McWilliams et al. Clin Genet 2002; 62:80-3
	<b>p.P2366R</b>	<b>This study</b>

Exon	Variant	Reference
44	p.A2367T	Sambuughin et al. Anesthesiology. 2001; 95:594-9
	p.G2375A	Ruffert et al. Anaesthesist 2002; 51:904-13
45	p.M2423K	Zhou et al. Neuromuscul Disord 2005; 15:676–743
	p.A2428T	Rueffert et al. Acta Anaesthesiol Scand 2002; 46:692-8
	p.D2431N	Sambuughin et al. Anesthesiology. 2001; 95:594-9
	p.G2434R	Keating et al. Hum Mol Genet 1994; 3:1855-8
	<b>p.R2435H</b>	Zhang et al. Nat Genet 1993; 5:46-50
	p.R2435L	Barone et al. J Med Genet 1999; 36:115-8
46	<b>p.R2452Q</b>	<b>This study</b>
	<b>p.R2452W</b>	Chamley et al. Br J Anaesth 2000; 84:500-4
	p.I2453T	Ruffert et al. Anaesthesist 2002; 51:904-13
	p.R2454H	Barone et al. J Med Genet 1999; 36:115-8
	p.R2454C	Brandt et al. Hum Mol Genet 1999; 8:2055-62
	<b>p.R2458H</b>	Manning et al. Hum Mutat 1998; 11:45-50
	p.R2458C	Manning et al. Hum Mutat 1998; 11:45-50
47	<b>p.P2496L</b>	<b>This study</b>
	<b>p.R2508C</b>	<b>This study</b>
	<b>p.R2508G</b>	<b>This study</b>
	<b>p.R2508H</b>	<b>This study</b>
48	<b>p.E2545D</b>	<b>This study</b>
50	p.R2676W	Guis et al. Arch Neurol 2004; 61:106-13
51	<b>p.D2730H</b>	<b>This study</b>
	p.G2733D	Sambuughin et al. Anesthesiology 2005; 102:515-21
53	p.S2776M	Zhou et al. Neuromuscul Disord 2005; 15:676–743

Exon	Variant	Reference
53	p.T2787S	Guis et al. Arch Neurol 2004; 61:106-13
54	<b>p.R2840W</b>	<b>This study</b>
57	p.R2939K	Jungbluth et al. Neuromuscul Disord 2005; 15:676–743
	p.R2939S	Zhou et al. Neuromuscul Disord 2005; 15:676–743
63	<b>p.R3119H</b>	<b>This study</b>
67	p.R3348H	Sambuughin et al. Anesthesiology 2005; 102:515-21
	<b>p.K3367R</b>	<b>This study</b>
68	p.S3446F	Zhou et al. Neuromuscul Disord 2005; 15:676–743
71	p.P3527S	Zhou et al. Neuromuscul Disord 2005; 15:676–743
73	<a href="#">p.L3606P</a>	<a href="#">Wu et al. Brain 2006 (in press).</a>
79	<a href="#">p.Q3756E</a>	Sambuughin et al. Anesthesiology 2005; 102:515-21
82	<b>p.V3840I</b>	<b>This study</b>
86	p.I3916M	Monnier et al. Anesthesiology 2002; 97:1067-74
89	<b>p.T4081M</b>	<b>This study</b>
90	p.N4119Y	Sambuughin et al. Anesthesiology 2005; 102:515-21
	p.R4136S	Galli et al. Cell Calcium 2002; 32:143-51
91	p.R4214_F4216del	Monnier et al. Hum Mol Genet 2000; 9:2599-608
	p.V4234L	Galli et al. Cell Calcium 2002; 32:143-51
	<b>p.E4283V</b>	<b>This study</b>
	<b>p.L4320_R4322dup</b>	<b>This study</b>
	p.R4326D	Zhou et al. Neuromuscul Disord 2005; 15:676–743
93	p.R4549Q	Kossugue et al. Neuromuscul Disord 2005; 15:676–743
94	<a href="#">p.L4568P</a>	<a href="#">Wu et al. Brain 2006 (in press).</a>
95	<a href="#">p.Y4631N</a>	<a href="#">Wu et al. Brain 2006 (in press).</a>

Exon	Variant	Reference
95	<a href="#">p.E4634K</a>	<a href="#">Wu et al. Brain 2006 (in press).</a>
	p.T4637A	Scacheri et al. Neurology 2000; 55:1689-96
	p.T4637I	Davis et al. Neuromuscul Disord 2003; 13:151-7
	<a href="#">p.G4638S</a>	<a href="#">Wu et al. Brain 2006 (in press).</a>
	p.G4638D	Davis et al. Neuromuscul Disord 2003; 13:151-7
	<b>p.R4645Q</b>	<b>This study</b>
	p.LS4647-8del	Monnier et al. Hum Mol Genet 2000; 9:2599-608
	p.H4651P	Davis et al. Neuromuscul Disord 2003; 13:151-7
	p.L4665P	Zhou et al. Neuromuscul Disord 2005; 15:676–743
96	p.P4668S	Oyamada et al. Jpn J Pharmacol 2002; 88:159-66
	p.F4684S	Monnier et al. Hum Mutat 2005; 26:413-25
	p.T4709M	Zhou et al. Neuromuscul Disord 2005; 15:676–743
97	p.K4724Q	Zhou et al. Neuromuscul Disord 2005; 15:676–743
98	p.Y4733D	Sambuughin et al. Anesthesiology 2005; 102:515-21
	p.R4737W	Galli et al. Cell Calcium 2002; 32:143-51
	p.R4737Q	Monnier et al. Hum Mutat 2005; 26:413-25
100	p.Y4792P	Monnier et al. Hum Mol Genet 2000; 9:2599-608
	p.Y4796C	
	p.F4808N	Davis et al. Neuromuscul Disord 2003; 13:151-7
	p.F4808del	Medne et al. Neuromuscul Disord 2005; 15:676–743
	p.L4814F	Shepherd et al. J Med Genet 2004; 41:e33
	p.L4824P	Shepherd et al. J Med Genet 2004; 41:e33
	p.R4825C	Monnier et al. Hum Mol Genet 2000; 9:2599-608
	p.T4826I	Brown et al. Hum Mol Genet 2000; 9:1515-24

Exon	Variant	Reference
101	<b>p.L4838V</b>	Oyamada et al. Jpn J Pharmacol 2002; 88:159-66
	p.A4846V	Kossugue et al. Neuromuscul Disord 2005; 15:676–743
	p.V4849I	Jungbluth et al. Neurology 2002; 59:284-7
	p.T4853I	Haugen et al. Tidsskr Nor Laegeforen 2005;125:2792-4
	<b>p.N4858D</b>	<a href="#">Wu et al. Brain 2006 (in press).</a>
	p.F4860del	Monnier et al. Hum Mol Genet 2001; 10:2581-92
	p.R4861H	Tilgen et al. Hum Mol Genet 2001; 10:2879-87
	p.R4861C	Davis et al. Neuromuscul Disord 2003; 13:151-7
	p.F4863_D4869del	Zorzato et al. Hum Mol Genet 2003; 12:379-88
	p.Y4864C	Zorzato et al. Hum Mol Genet 2003; 12:379-88
102	p.K4876R	Sambuughin et al. Anesthesiology 2005; 102:515-21
	p.M4880T	Sambuughin et al. Anesthesiology 2005; 102:515-21
	p.G4891R	Tilgen et al. Hum Mol Genet 2001; 10:2879-87
	p.R4893W	Monnier et al. Hum Mol Genet 2001; 10:2581-92
	p.R4893Q	Davis et al. Neuromuscul Disord 2003; 13:151-7
	<b>p.R4893P</b>	<a href="#">Wu et al. Brain 2006 (in press).</a>
	<b>p.A4894T</b>	<b>This study</b>
	p.G4897V	Kossugue et al. Neuromuscul Disord 2005; 15:676–743

Exon	Variant	Reference
102	p.I4898T	Lynch et al. Proc Natl Acad Sci U S A 1999; 96:4164-9
	<b>p.G4899E</b>	<a href="#">Wu et al. Brain 2006 (in press).</a>
	p.G4899R	Tilgen et al. Hum Mol Genet 2001; 10:2879-87
	p.A4906V	Tilgen et al. Hum Mol Genet 2001; 10:2879-87
	p.R4914G	Monnier et al. Hum Mol Genet 2001; 10:2581-92
	p.R4914Q	Zhou et al. Neuromuscul Disord 2005; 15:676–743
	p.R4914T	Davis et al. Neuromuscul Disord 2003; 13:151-7
	<b>p.T4920N</b>	<a href="#">Wu et al. Brain 2006 (in press).</a>
	<b>p.F4921T</b>	<a href="#">Wu et al. Brain 2006 (in press).</a>
	<b>p.F4921S</b>	<a href="#">Wu et al. Brain 2006 (in press).</a>
103	p.V4927_I4928del	Davis et al. Neuromuscul Disord 2003; 13:151-7
	p.I4938M	Shepherd et al. J Med Genet 2004; 41:e33
	p.D4939E	Shepherd et al. J Med Genet 2004; 41:e33
	p.A4940T	Davis et al. Neuromuscul Disord 2003; 13:151-7
104	p.G4942V	Galli et al. Cell Calcium 2002; 32:143-51
	<b>p.F4960Y</b>	<b>This study</b>
	p.P4973L	Monnier et al. Anesthesiology 2002; 97:1067-74