Title: Spastic Paraplegia Type 11 *GeneReview* Table 3

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Note: The following information is provided by the authors listed above and has not been reviewed by GeneReviews staff.

Table 3. *SPG11* Pathologic Allelic Variants [1]

Exons or Introns	DNA Nucleotide change	Protein Amino acid change of effect on mRNA	Key reference
1	c.118C>T	p.Q40X	Stevanin et al 2007; Stevanin et al 2008
2	c.267G>A	p.W89X	Hehr et al 2007, Paisan-Ruiz et al 2008b
2	c.268G>T	p.E90X	Denora et al, 2008
2	c.349G>T	p.E117X	Crimella et al 2009
2	c.359delT	L120X	Samaranch al 2008
2	c.398delG	p.C133LfsX15	Paisan-Ruiz et al 2008b
2	c.408_428del21	p.E136_I142del	Denora et al, 2008
Intron2	c.442+1G>C	r.258_442del185	Hehr et al 2007
3	c.529_533delATATT	p.I177_F178>S177fsX178	Stevanin et al 2007; Denora et al 2008
3	c.642delT	p.F214LfsX3	Orlen et al 2009
3	c.654_655delinsG	p.S218RfsX219	Liao et al 2008
4	c.704_705delAT	p.H235RfsX246	Stevanin et al 2008
4	c.733_734delAT	p.M245VfsX246	DelBo et al 2007; Hehr et al 2007; Stevanin et al 2007; Stevanin et al 2008; Denora et al, 2008; Boukhris et al 2008; Liao et al 2008
Intron4	c.869+1G>A	r.?	Stevanin et al 2008
6	c.1203delA	p.K401KfsX415	Stevanin et al 2007; Denora et al 2008
6	c.1235C>G	p.S412X	Stevanin et al 2008, Samaranch et al 2008
6	c.1282A>T	p.K428X	Stevanin et al 2008
6	insA	p.I450NfsX475	Paisan-Ruiz et al 2008
Intron6	c.1457-2A>G	r.? Skipping of exon 7	Hehr et al 2007

7	c.1471_1472delCT	p.L491DfsX556	Stevanin et al 2008
7		p.Q498X	Abdel-Aleem et al 2011
7	c.1549_1550delCT	p.L517LfsX556	Stevanin et al 2008
7	c.1550_1551delTT	L517fsX556	Samaranch al 2008
8	c.1668delT	p. F556LfsX577	Stevanin et al 2008
8	c.1679C>G	p.S560X	Denora et al, 2008
8	c.1697_1712del16insTACTCCCAT	p.D566VfsX595	Denora et al, 2008
Intron 8	c.1735+3_+6delAAGT	r. ?	Crimella et al 2009
9	c.1837_18388insA	p.L613HfsX619	Paisan-Ruiz et al 2008
9	c.1845_1846delGT	p.S616fs618X	Hehr et al 2007
10	c.1951C>T	p.R651X	Stevanin et al 2008; Hehr et al 2007
11	c.2146C>T	p.Q716X	Southgate et al 2010
11	c.2163_2164insT	p.I722YfsX731	Liao et al 2008
11	c.2198T>G	p.L733X	Stevanin et al 2007
Intron 12	c.2316+1G>A	r.?	Stevanin et al 2008
13	c.2355_2356del2	p.K785SfsX796	Denora et al, 2008
13	c.2444G>T	p.R815M, r.?	Denora et al, 2008
Intron13	c.2444+1G>C	r.?	Denora et al, 2008
14	c.2472insT	p.K825fs837X	Hehr et al 2007
14	c.2608A>G	r. ?	Pippucci et al, 2009
15	c.2697G>A	p.W899X	Denora et al, 2008
15	c.2716delC	p.Q906SfsX920	Denora et al, 2008
15	c.2833A>G	r.2834+1_2834+65ins, p.R945GfsX5	Stevanin et al 2008; Denora et al 2008 ; Crimella et al 2009
intron15	c.2834+1G>T	r. ?	Paizan-Ruiz et al 2008b
16	c.2842_2843insG	p.V948GfsX953	Stevanin et al 2007
16	c.2849_2850insT	p.L950FfsX953	Denora et al, 2008
17	c.3075_3076insA	p.R1026fs1029X	Hehr et al 2007; Denora et al, 2008
17	c.3145_3146insCA	p.T1048fs1053X	Hehr et al 2007
Intron18	c.3291+1G>T	r. ?	Kim et al, 2009
21	c.3602_3603delAT	p.Y1201LfsX4	Southgate et al 2010
21	c.3664_3665insT	p.K1222IfsX1236	Denora et al, 2008

22	c.3719_3720delTA	p.I1240VfsX263	Liao et al 2008
22	c.3741_3742insA	p.P1248TfsX1264	Denora et al, 2008
24	c.4046T>A	p.F1349I	Denora et al 2008
25	c.4307_4308delAA	p.Q1436RfsX1442	Stevanin et al 2008; Denora et al 2008, Zhang et al 2008
26	c.4461_4462delGT	p.Cys1487fsX	Lee et al 2008
27	c.4668T>A	p.Y1556X	Liao et al 2008
28	c.4846C>T	p.Q1616X	Southgate et al 2010
30	c.5255delT	p.F1752fs1765X	Hehr et al 2007; Denora et al 2008
30	insA	p.E1819AfsX1828	Paisan-Ruiz et al 2008
30	c.5399_5407delAGATinsTGGAGGAG	p.Q1800LfsX1830	Paisan-Ruiz et al 2008
30	c.5410_5411delTG	p.C1804PfsX25	Kim et al 2009
30	c.5470C>T	p.R1824X	Denora et al, 2008
30	c.5532_5533delCA	p.S1844SfsX1857	Stevanin et al 2008
30	c.5623C>T	p.Q1875X	Paizan-Ruiz et al 2008b
30	c.5703delT	p.F1901fsX1950	Samaranch al 2008
30	c.5769delT	p.S1923RfsX1950	Stevanin et al 2008, Paisan-Ruiz et al, 2008
30	c.5798delC	p.A1933fs1951X	Hehr et al 2007
31-34	c.5867-3237_6478-451del8323	p.T1956SfsX18	Bauer et al 2008
31	c.5870C>G	p.S1957X	Denora et al 2008
31-34	c.5898+5493_6509-491del	p.T1966fsX1968	Denora et al 2008
31	c.5970C>G	p.Y1990X	Denora et al 2008
31	c.5974C>T	p.R1992X	Stevanin et al 2007
31	c.5977C>T	c.5977C>T/p.Q1993X	Zhang et al 2008, Liao et al 2008
31	c.5986_5987insT	p.C1996LfsX1999	Stevanin et al 2008; Denora et al 2008
31	c.5985delCTGT	p.L1997MfsX2056	Paisan-Ruiz et al 2008
31	c.5987_5990dupCTCT	p.C1996fsX1999	Denora et al 2008
31	c.5989_5992delCTGT	p.L1997_Y1998>M1997fsX2056	Stevanin et al 2008

31	c.5992insT	p.Y1998fsX1999	Denora et al 2008
32	c.6091C>T	p.R2031X	Lee et al, 2008 ; Denora et al 2008
32	c.6100C>T	p.R2034X	Stevanin et al 2007, Stevanin et al 2008, Denora et al, 2008, Boukhris et al 2008, Samaranch et al 2008
32	c.6157G>A	p.V2053M	Crimella et al 2009
Intron 32	c.6206-1G>C	r. ?	Orlen et al 2009
34	c.6451delG	p. A2151PfsX2172	Stevanin et al 2007
Intron 34	c.6477+4 A>G	r.?	Stevanin et al 2008
36	c.6737_6740delTTGA	p.I2246_E2247>S2246fsX2260	Stevanin et al 2008
36	c.6739_6742delGAGT	p.E2247_S2248>L2247fsX2260	Stevanin et al 2008
36	c.6754+4insTG	r. ?	Paizan-Ruiz et al 2008b
37-39	g.96677_99386del2710, r.6755_7151del397	p.E2252DfsX15	Crimella et al 2009
37	c.6790_6791insC	p.L2264PfsX2339	Liao et al 2008
37	c.6832_6833delAG	p.S2278LfsX2338	Stevanin et al 2008
38	c.6856C>T	p.R2286X	Denora et al 2008
38	c.6898_6899delCT	p.L2300AfsX2338	Liao et al 2008
Intron38	c.7000-3_-2insGGA	p.A2334EfsX7	Crimella et al 2009
39	c.7023C>A	p.Y2341X	Denora et al 2008
39	c.7029_7030insT	p.V2344CfsX2349	Stevanin et al 2007
39	c.7088_7089insATTA	p.Y2363X	Liao et al 2008
39	c.7101_7102insT	p.K2368X	Liao et al 2008
Intron39	c.7151+4_7151+7delAGTA	p.K2384fsX2386	Liao et al 2008
Intron39	c.7152-1G>C	r. ?	Orlen et al 2009
40	c.7156_7157insAAAC	p.K2386QfsX2393	Paizan-Ruiz et al 2008b

1. SPG11 Reference sequences: NM_025137.3 and NP_079413.3

2. A designation of “r.?” indicates that the RNA has not been analyzed and an effect is expected but difficult to predict [www.hgvs.org].

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