Note: The following information is provided by the author(s) 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 or effect on mRNA (when known) [2]	Key References
1	c.118C>T	p.Q40X	Stevanin, Santorelli et al 2007; Stevanin Azzedine et al 2008
2	c.267G>A	p.W89X	Hehr et al 2007
Intron2	c.442+1G>C	r.?	Hehr et al 2007
3	c.529_533delATATT	p.I177_F178>S177fsX178	Stevanin, Santorelli et al 2007
4	c.704_705delAT	p.H235RfsX246	Stevanin, Azzedine et al 2008
4	c.733_734delAT	p.M245VfsX246	DelBo et al 2007; Hehr et al 2007; Stevanin, Santorelli et al 2007; Stevanin, Azzedine et al 2008
Intron4	c.869+1G>A	r.?	Stevanin, Azzedine et al 2008
6	c.1203delA,	p.K401KfsX415	Stevanin, Santorelli et al 2007
6	c.1235C>G	p.S412X	Stevanin, Azzedine et al 2008
6	c.1282A>T	p.K428X	Stevanin, Azzedine et al 2008
Intron6	c.1757-2A>G	Skipping of exon 7	Hehr et al 2007
7	c.1471_1472delCT	p.L491DfsX556	Stevanin, Azzedine et al 2008
7	c.1549_1550delCT	p.L517LfsX556	Stevanin, Azzedine et al 2008
8	c,1668delT,	p. F556LfsX577	Stevanin, Azzedine et al 2008
9	c.1845_1846delGT	p.S616fs618X	Hehr et al 2007
10	c.1951C>T	p.R651X	Stevanin, Azzedine et al 2008; Hehr et al 2007
11	c.2198T>G	p.L733X	Stevanin, Santorelli et al 2007
Intron 12	c.2316+1G>A	r.?	Stevanin, Azzedine et al 2008
14	c.2472insT	p.K825fs837X	Hehr et al 2007
15	c.2833A>G	r.2834+1_2834+65ins, p.R945GfsX5	Stevanin, Azzedine et al 2008
16	c.2842_2843insG	p.V948GfsX953	Stevanin, Santorelli et al 2007
17	c.3076insA	p.R1026fs1029X	Hehr et al 2007
17	c.3145_3146insCA	p.T1048fs1053X	Hehr et al 2007
25	c.4307_4308delAA	p.Q1436RfsX1442	Stevanin, Azzedine et al 2008
30	c.5255delT	p.F1752fs1765X	Hehr et al 2007
30	c.5532_5533delCA	p.S1844SfsX1857	Stevanin, Azzedine et al 2008
30	c.5769delT	p.S1923RfsX1950	Stevanin, Azzedine et al 2008
30	c.5798delC	p.A1933fs1951X	Hehr et al 2007
31	c.5974C>T	p.R1992X	Stevanin, Santorelli et al 2007
31	c.5986_5987insT	p.C1996LfsX1999	Stevanin, Azzedine et al 2008

Exons or introns	DNA Nucleotide Change	Protein Amino Acid Change or effect on mRNA (when known) [2]	Key References
31	c.5989_5992delCTGT	p.L1997_Y1998>M1997fsX2056	Stevanin, Azzedine et al 2008
32	c.6100C>T	p.R2034X	Stevanin, Santorelli et al 2007; Stevanin, Azzedine et al 2008
34	c.6451delG	p. A2151PfsX2172	Stevanin, Santorelli et al 2007
Intron 34	c.6477+4 A>G	r.?	Stevanin, Azzedine et al 2008
36	c.6737_6740delTTGA	p.I2246_E2247>S2246fsX2260	Stevanin, Azzedine et al 2008
36	c.6739_6742delGAGT	p.E2247_S2248>L2247fsX2260	Stevanin, Azzedine et al 2008
37	c.6832_6833delAG	p.S2278LfsX2338	Stevanin, Azzedine et al 2008
39	c.7029_7030insT	p.V2344CfsX2349	Stevanin, Santorelli et al 2007

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)

References

DelBo R, DiFonzo A, Ghezzi S, Locatelli F, Stevanin G, Costa A, Corti S, Bresolin N, Comi GP (2007) SPG11: a consistent clinical phenotype in a family with homozygous Spatacsin truncating mutation. *Neurogenetics* 8:301-5

Hehr U, Bauer P, Winner B, Schule R, Olmez A, Koehler W, Uyanik G, Engel A, Lenz D, Seibel A, Hehr A, Ploetz S, Gamez J, Rolfs A, Weis J, Ringer TM, Bonin M, Schuierer G, Marienhagen J, Bogdahn U, Weber BH, Topaloglu H, Schols L, Riess O, Winkler J (2007) Long-term course and mutational spectrum of spatacsin-linked spastic paraplegia. *Ann Neurol* 62:656-65

Stevanin G, Azzedine H, Denora P, Boukhris A, Tazir M, Lossos A, Rosa AL, Lerer I, Hamri A, Alegria P, Loureiro J, Tada M, Hannequin D, Anheim M, Goizet C, Gonzalez-Martinez V, Le Ber I, Forlani S, Iwabuchi K, Meiner V, Uyanik G, Erichsen AK, Feki I, Pasquier F, Belarbi S, Cruz VT, Depienne C, Truchetto J, Garrigues G, Tallaksen C, Tranchant C, Nishizawa M, Vale J, Coutinho P, Santorelli FM, Mhiri C, Brice A, Durr A (2008) Mutations in SPG11 are frequent in autosomal recessive spastic paraplegia with thin corpus callosum, cognitive decline and lower motor neuron degeneration. *Brain* 131:772-84

Stevanin G, Santorelli FM, Azzedine H, Coutinho P, Chomilier J, Denora PS, Martin E, Ouvrard-Hernandez AM, Tessa A, Bouslam N, Lossos A, Charles P, Loureiro JL, Elleuch N, Confavreux C, Cruz VT, Ruberg M, Leguern E, Grid D, Tazir M, Fontaine B, Filla A, Bertini E, Durr A, Brice A (2007) Mutations in SPG11, encoding spatacsin, are a major cause of spastic paraplegia with thin corpus callosum. *Nat Genet* 39:366-72