Title: Spastic Paraplegia 7 GeneReview Table 4

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

Table 4. *SPG7* Pathologic Allelic Variants

***SPG7* Location, Exon**	**DNA Nucleotide Change**	**Protein Amino Acid Change**	**Reference**	**Reference Sequences**
1	1A>T	initiation	Elleuch et al 2006	NM_003119.2 NP_003110.1
1	28G>A	Ala10Ser	Wilkinson et al 2004	
2	233T>A	Leu78X non sense	Arnoldi et al 2008	
2	244-246delACA	Gln82del	Elleuch et al 2006	
5	698T>C	Leu233Pro	Arnoldi et al 2008	
6	784del2	Frameshift, truncated protein	Casari et al 1998	
6	850_-851 delTTinsC	Phe284ProfsX44	Elleuch et al 2006	
8	1045G>A	Gly349Ser	Bonn et al 2010	
8	1047insC	Frameshift, truncated protein	Tzoulis et al 2008	
8	1057_-1085del29	Frameshift 353-384X385	Wilkinson et al 2004	
11-13	1447-1778 del 331	Glu484_Lys593 del119	Arnoldi et al 2008	
11	1450-1458del 9	Glu,Arg,Arg484-486del	McDermott et al 2001	
11	1519 C>T	Glu507X	Elleuch et al 2006	
Intron 11	155211 G>T	splite site	Warnecke et al 2010	
12	1616delC	Val540fs	Arnoldi et al 2008	
12	1636G>A	Glu546Lys	Arnoldi et al 2008	
13	1749G>C	Trp583Cys	Bonn et al 2010	
13	1715C>T	Ala572Val	Wilkinson et al 2004	
12-17	del 9,5kb	Large deletion	Casari et al 1998	
13	1729G>A	Gly577Ser	Wilkinson et al 2004	
13	1742-1744del3	Val581del	Elleuch et al 2006	
14	1904C>T	Ser635Leu	Elleuch et al 2006	
15	1948G>C	Asp650His	Elleuch et al 2006	
15	2026T>C	Phe676Leu	Wilkinson et al 2004	
15	2075G>C	Ser692Thr	Warnecke et al 2007	
17	2191G>A	Ala731Thr	Arnoldi et al 2008	
17	2216dupA	Asn739fs	Arnoldi et al 2008	
17	2228 Ins A	Frameshift, stop	Casari et al 1998	

See Quick Reference for an explanation of nomenclature. *GeneReviews* follows the standard naming conventions of the Human Genome Variation Society (www.hgvs.org).

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