Title: *ALS2*-Related Disorders *GeneReview,* Table 3

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Note: The following information was provided by the original authors (Bertini ES, Eymard-Pierre E, Boespflug-Tanguy O, Cleveland DW, Yamanaka K) and was not reviewed by *GeneReviews* staff.

Table 3. Selected *ALS2* Pathogenic Allelic Variants

DNA Nucleotide Change (Alias [1])	Protein Amino Acid Change	Reference Sequences [2]	Pheno-type	References
c.138delA (A261del)	p.Ala47ProfsTer4	NM_020919.2 NP_065970.2	JALS	Hadano et al [2001], Yang et al [2001]
c.470G>A	p.Cys157Tyr		IAHSP	Eymard-Pierre et al [2006]
c.553delA	p.Thr185fsTer5		JALS	Kress et al [2005]
c.1007_1008delTA	p.Ile336ThrfsTer5		IAHSP	Eymard-Pierre et al [2002]
c.1425_1426delAG (1548delAG)	p.Glu476GlyfsTer71		JPLS	Hadano et al [2001], Yang et al [2001]
c.1472_1481delTTTCCCCCAG	p.Val491GlyfsTer3		IAHSP	Eymard-Pierre et al [2002]
c.1619G>A	p.Gly540Glu		JPLS	Panzeri et al [2006]
C;.1825_1826insCAGTG/ c.3529G>T	p.Glu609fsTer9/ p.Gly1177Ter		IAHSP	Sztriha et al [2008]
c.1867_1868delCT	p.Leu623ValfsTer24		JPLS	Yang et al [2001]
c.2143C>T	p.Gln715Ter		IAHSP	Verschuuren-Bemelmans et al [2008]
IVS9-2A>T	p.Glu724fsTer32		IAHSP	Herzfeld et al [2008]
c.2537_2538delAT (2660delAT)	p.Asn845IlefsTer13		IAHSP	Eymard-Pierre et al [2002]
c.2980-2A>G	p.993fsTer7		JPLS	Mintchev et al [2009]
c.2992C>T (C3115T)	p.Arg998Ter		IAHSP	Devon et al [2003]
c.3565delG / IVS22+5G>C	p.Val1189TrpfsTer19/ p.Gly1172GlufsTer29		JALS	Shirakawa et al [2009]
c.3619delA (3742delA)	p.Met1206Ter		IAHSP	Eymard-Pierre et al [2002]
c.4721delT (4844delT)	p.Val1574AlafsTer44		IAHSP	Gros-Louis et al [2003]

Note on variant classification: Variants listed in the table have been provided by the author(s). *GeneReviews* staff have not independently verified the classification of variants.

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

1. Variant designations that do not conform to current naming conventions

2. Reference sequence (www.ncbi.nlm.nih.gov/Genbank/index.html)

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