

Title: Adult-Onset Leukoencephalopathy with Axonal Spheroids and Pigmented Glia *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. Selected *CSF1R* Pathogenic Allelic Variants

DNA Nucleotide Change (Alias ¹)	Protein Amino Acid Change	Reference Sequences
c.1754-2A>G	p.Gly585_Lys619delinsAla ²	NM_005211.3 NP_005202.2
c.1766G>A	p.Gly589Glu ²	
c.1897G>A	p.Glu663Lys ²	
c.2297T>C	p.Met766Thr ²	
c.2308G>C	p.Ala770Pro ²	
c.2320-2A>G	p.Cys774_Asp814del ²	
c.2324T>A	p.Ile775Asn ²	
c.2381T>C	p.Ile794Thr ^{2, 3, 4}	
c.2442+5G>C	p.Cys774_Asp814delinsGlnGlyLeuGlnSerHisVal GlyProSerLeuProSerSerSerProGlnAlaGln ²	
c.2546_2548delTCT	p.Phe849del ²	
c.2546T>C	p.Phe849Ser ²	
c.2603T>C	p.Leu868Pro ²	
c.2624T>C	p.Met875Thr ²	
c.2632C>A	p.Pro878Thr ²	
c.2345G>A	p.Arg782His ⁵	
c.2329C>T	p.Arg777Trp ³	
c.1958G>A	p.Cys653Tyr ³	
c.2483T>C (2775T>C)	p.Phe828Ser ⁴	
c.1699delA	p.Thr567fsTer44 ⁶	
c.1889T>G	p.Leu630Arg ⁶	
c.2329C>T	p.Arg777Trp ⁶	
c.2330G>A	p.Arg777Gln ^{6, 8}	
c.2450T>C	p.Leu817Pro ⁶	
c.2480T>C	p.Ile827Thr ⁶	
c.2541G>C	p.Glu847Asp ⁶	
c.2566T>C	p.Tyr856His ⁶	
c.2701C>T	p.Pro901Ser ⁶	
c.2655-2A>G	Not available ⁶	
c.2342C>T	p.Ala781Val ⁷	

DNA Nucleotide Change (Alias ¹)	Protein Amino Acid Change	Reference Sequences
c.2528T>A	p.Ile843Asn ⁷	
c.2629C>T	p.Gln877Ter ⁷	
c.2512G>C	p.Val838Leu ⁷	
c.2527A>T	p.Ile843Phe ⁹	
c.1957T>C	p.Cys653Arg ⁹	
c.2717T>C	p.Ile906Thr ⁹	
c.2378A>C	p.Lys793Thr ¹⁰	
c.2467C>T	p.Ala823Val ¹¹	
c.1745T>C	p.Leu582Pro ¹²	
c.2350G>A	p.Val784Met ¹³	
c.2562T>A	p.Asn854Lys ¹⁴	
c.2294G>A	p.Gly765Asp ¹⁵	
c.2342C>A	p.Ala781Glu ¹⁵	
c.2442+1G>T	Not available ¹⁵	
c.2060_2061insT	p.Ser688GlufsTer1 ¹⁵	

Note on variant classification: Variants listed in the table have been provided by the authors. *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 designation that does not conform to current naming conventions

2. Rademakers et al [2011]

3. Mitsui et al [2012]

4. Kleinfeld et al [2012]

5. Kinoshita et al [2012]

6. Guerreiro et al [2013]

7. Karle et al [2013]

8. Inui et al [2013]

9. Battisti et al [2014]

10. Kondo et al [2013]

11. Terasawa et al [2013]

12. Schuberth et al [2014]

13. La Piana et al [2014]

14. Sundal et al [2014]

15. Konno et al [2014]

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