

Title: Geleophysic Dysplasia *GeneReview* Table 3

Authors: LeGoff C, Cormier-Daire V

Date: April 2012

Note: The following information is provided by the author(s) listed above and has not been reviewed by *GeneReviews* staff.

Table 3. Selected *ADAMTSL2* Pathologic Allelic Variants

DNA Nucleotide Change	Protein Amino Acid Change
c.2717C>T	p.Pro906Leu
c.475C>T	p.Arg159Trp
c.215G>A	p.Arg72Gln
c.150G>T	p.Trp50Cys
c.493G>A	p.Ala165Thr
c.1273C>T	p.Arg425X
c.511T>C	p.Cys171Arg
c.661C>T	p.Arg221Cys
c.715G>A	p.Ala239Thr
c.1777C>T	p.Arg593Cys
c.1904C>T	p.Ser635Leu

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