

Title: *ADAMTSL4*-Related Eye Disorders *GeneReview* Table 3

Authors: Rødahl E, Christensen AE, Fiskerstrand T, Knappskog PM, Boman H

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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. Published *ADAMTSL4* Allelic Variants per December 2011

Class of Variant Allele	DNA Nucleotide Change	Protein Amino Acid Change	Reference Sequences
Pathologic	c.79-1G>A	p.?	NM_019032.4 NP_061905.2
	c.767_786del	p.Gln256ProfsX38	
	c.826_836del	p.Arg276SerfsX21	
	c.926G>A	p.Arg309Gln	
	c.1785T>G	p.Tyr595X	
	c.1960C>T	p.Pro654Ser	
	c.2008C>T	p.Arg670X	
	c.3153C>A	p.Tyr1051X	
	c.3161A>G	p.Tyr1054Cys	

Ahram et al [2009], Aragon-Martin et al [2010], Greene et al [2010], Christensen et al [2010], Neuhaan et al [2011]

References

Ahram D, Sato TS, Kohilan A, Tayeh M, Chen S, Leal S, Al-Salem M, El-Shanti H (2009) A homozygous mutation in *ADAMTSL4* causes autosomal-recessive isolated ectopia lentis. *Am J Hum Genet* 84:274-278.

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