Title: CARASIL *GeneReview* Table 3

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

Table 3*. HTRA1* Allelic Variants

Variant Classification	DNA Nucleotide Change (Alias [1])	Protein Amino Acid Change	Reference Sequences
Benign	c.102C>T (230C>T)	p.Ala34Ala	NM_002775.4 NP_002766.1
	c.108G>C (236G>C)	p.Gly36Gly	
	c.108G>T (236G>T)	p.Gly36Gly	
Pathogenic	c.754G>A	p.Ala252Thr	Yanagawa et al [2002]
	c.821G>A	p.Arg274Gln	Nishimoto et al [2011]
	c.854C>T	p.Pro285Leu	Chen et al [2013]
	c.883G>A	p.Gly295Arg	Mendioroz et al [2010]
	c.889G>A	p.Val297Met	Hara et al [2009]
	c.904C>T	p.Arg302Ter	Hara et al [2009]
	c.1091T>C	p.Leu364Pro	Wang et al [2012]
	c.1108C>T	p.Arg370Ter	Hara et al [2009], Bayrakli et al [2014]
	c.[126delG];[961G>A]	p.[Glu42fs];[Ala321Thr]	Bianchi et al [2014]

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

References

Bayrakli F, Balaban H, Gurelik M, Hizmetli S, Topaktas S. Mutation in the HTRA1 gene in a patient with degenerated spine as a component of CARASIL syndrome. Turk Neurosurg. 2014;24:67-9.

Bianchi S, Di Palma C, Gallus GN, Taglia I, Poggiani A, Rosini F. et al. Two novel HTRA1 mutations in a European CARASIL patient. Neurology. 2014;82:898–900.

Chen Y, He Z, Meng S, Li L, Yang H, Zhang X. A novel mutation of the high-temperature requirement A serine peptidase 1 (HTRA1) gene in a Chinese family with cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy (CARASIL). J Int Med Res. 2013;41:1445–55.

Hara K, Shiga A, Fukutake T, Nozaki H, Miyashita A, Yokoseki A, Kawata H, Koyama A, Arima K, Takahashi T, Ikeda M, Shiota H, Tamura M, Shimoe Y, Hirayama M, Arisato T, Yanagawa S, Tanaka A, Nakano I, Ikeda S, Yoshida Y, Yamamoto T, Ikeuchi T, Kuwano R, Nishizawa M, Tsuji S, Onodera O. Association of HTRA1 mutations and familial ischemic cerebral small-vessel disease. N Engl J Med. 2009;360:1729–39.

Mendioroz M, Fernández-Cadenas I, Del Río-Espinola A, Rovira A, Solé E, Fernández-Figueras MT, García-Patos V, Sastre-Garriga J, Domingues-Montanari S, Alvarez-Sabín J, Montaner J. A missense HTRA1 mutation expands CARASIL syndrome to the Caucasian population. Neurology. 2010;75:2033-5.

Nishimoto Y, Shibata M, Nihonmatsu M, Nozaki H, Shiga A, Shirata A, Yamane K, Kosakai A, Takahashi K, Nishizawa M, Onodera O, Suzuki N. A novel mutation in the HTRA1 gene causes CARASIL without alopecia. Neurology. 2011;76:1353-5.

Wang XL, Li CF, Guo HW, Cao BZ. A novel mutation in the HTRA1 gene identified in Chinese CARASIL pedigree. CNS Neurosci Ther. 2012;18:867-9.

Yanagawa S, Ito N, Arima K, Ikeda S (2002) Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy. Neurology. 58:817-20.