Title: Ataxia-Telangiectasia *GeneReview* – Table 3

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Updated: October 2016

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

Founder variants have been described in many populations.

Table 3. *ATM* Pathogenic Allelic Variants in Ethnic Populations

Ethnicity	DNA Nucleotide Change (Alias [1])	Predicted Protein Change	Allele Frequency (%)
African American	c.2851-10T>G (IVS16-10T>G)	--	-
	c.2810_2811insCTAG	p.Glu937AspfsTer2	-
	c.7327C>T	p.Arg2443Ter	-
	c.7926A>C	p.Arg2642Ser	-
Amish	c.1564_1565delGA (1563delAG)	p.Glu522IlefsTer43	>95 [5]
Costa Rican	c.5908C>T	p.Gln1970Ter	56
	(IVS63del17kb)	--	7
	c.7449G>A	p.Trp2483Ter	12
	c.4507C>T	p.Gln1503Ter	12
	c.8264_8268del5 (8264del5)	p.Tyr2755CysfsTer12	4
	c.1120C>T	p.Gln374Ter	2
Iranian	c.4852C>T	p.Arg1618Ter	<5%
	c.8201_8212del11ins6 (8201del11ins6)	--	
Italian	c.7517_c.7520del4	p.Cys252AsnfsTer2	16
	c.3576G>A	(deletion of exon 26)	12 [5]
	c.3894dupT (3894insT)	p.Ala1299CysfsTer3	Sardinia (>95%)
Japanese	c.7883_7887del5 (7883del5)	p.Ile2629SerfsTer25	-
	c.5390+2T>C (IVS33+2T>C)	--	-
North African Jewish	c.103C>T	p.Arg35Ter	>99
Norwegian	c.3245_3247delATCinsTGAT (3245ATC>TGAT)	p.His1082LeufsTer14	55

Ethnicity	DNA Nucleotide Change (Alias [1])	Predicted Protein Change	Allele Frequency (%)
Polish	c.8313-2A>C (IVS53-2A>C(del159nt)) [2]	--	15
	c.6095G>A	(deletion of exon 43)	8
	c.7010_7011del (7010delGT)	p.Cys2337SerfsTer35	6
	c.5932G>T (5932G>T(del88))	p.Glu1978Ter	10 [3]
	c.1564_1565delGA (1563delAG)	p.Glu522IlefsTer43	6 [5]
Turkish	c.3576G>A		39 [5]
	c.5763-1050A>G (5762ins137)	See Table 2	18 [4]
	c.7637_7645del9 (7637del9)	p.Arg2547_Ser2549del	15 [5]
United Kingdom [6]	c.2T>C	p.Met1Val	3
	c.1564_1565delGA (c.1563delAG)	p.Glu522IlefsTer43	5 [5]
	c.2249ins9 [1]		5
	c.2284_2285delCT	p.Leu762ValfsTer2	3
	c.2639del200nt	Del exon 20	8
	c.3802delG	p.Val1268Ter	5
	c.5763-1050A>G (5762G>Ains137)	See Table 2	18 [4]
	c.6404_6405insTT	p.Arg2136Ter	3
	c.7271T>G	p.Val2424Gly (kinase dead)	3
	c.7638_7646del9 (7636del9)	p.Arg2547_Ser2549del delSRI	15
	c.8786+1G>A (c.8787ins14)		5
Utah Mormon	c.5395-12A>G (IVS32-12A>G)	--	--
	8494C>T	p.Arg2832Cys	--
	c.9372+1G>A (IVS62+1G>A)	--	--

See Table 2 for reference sequences.

1. Variant designation that does not conform to current naming conventions; based on previously published mutations

2. Mitui et al [2005]

3. Also found in Mennonite and Russian persons with A-T

4. Milder phenotype?

5. Widely disseminated

6. Stankovic et al [1998]

References

Mitui M, Bernatowska E, Pietrucha B, Piotrowska-Jastrzebska J, Eng L, Nahas S, Teraoka S, Sholty G, Purayidom A, Concannon P, Gatti RA (2005) ATM gene founder haplotypes and associated mutations in Polish families with ataxia-telangiectasia. Ann Hum Genet. 69:657-64.

Stankovic T, Kidd AM, Sutcliffe A, McGuire GM, Robinson P, Weber P, Bedenham T, Bradwell AR, Easton DF, Lennox GG, Haites N, Byrd PJ, Taylor AM (1998) ATM mutations and phenotypes in ataxia-telangiectasia families in the British Isles: expression of mutant ATM and the risk of leukemia, lymphoma, and breast cancer. Am J Hum Genet 62:334-45