

Supplement Table 1: Known pathogenic variants in *DNAAF5*

Variant	Location*	Inheritance	Reference	ClinVar ID
c.2394C>T (p.Arg792Ter)	HEAT 10	Ter, homozygous	Unpublished	
c.2384T>C (p.Leu795Pro)	HEAT 10	Missense, Homozygous	PMID:23040496	39684
c.2374C>T (p.Arg792Ter)	HEAT 10	Ter, unknown inheritance	Unpublished	1454678
c.2432-1G>C	HEAT 10	splice mutant, Homozygous	PMID:25232951	
c.2353_2352AG, (p.Ser785fs)	HEAT 10	FS, Homozygous	PMID:29358401	241202
c.2346C>G (p.Tyr782Ter)	HEAT 10	Ter, unknown inheritance	Not reported	454860
c.2108_2114delinsCCACCCTGGGT (p.Met703fs)	HEAT 8	FS, compound heterozygous	Unpublished‡	454858
c.2083-1G>C		homozygous	Unpublished	
c.2013_2028delinsATGGCC (p.Ala672fs)	HEAT 8	FS, unknown inheritance	Unpublished	649289
c.1784-1G>T		splice acceptor, unknown inheritance	Unpublished	454854
c.1777_1778del (p.Gln593fs)	HEAT 5-6	FS, unknown inheritance	Unpublished	1453546
c.1731G>T (p.Trp577Cys)	HEAT 6-7	Missense, compound heterozygous	Unpublished‡	
c.1499G>T (p.C500F)	HEAT 5-6	1.Missense, homozygous 2.compound heterozygous	PMID:29358401 PMID:29363216†	
c.1142_1147delAGCTGC (p.Gln381_Leu382del)	HEAT6	In-frame deletion, homozygous	Unpublished	
c.948G>A (p.Trp316Ter)	HEAT 4	nonsense, unknown inheritance	Unpublished	663589
c.926G>A (p.Trp309Ter)	HEAT 4	nonsense, unknown inheritance	Unpublished	651485
c.943C>T (p.Gln315Ter)	HEAT 4	Nonsense, unknown inheritance	Unpublished	1432731
c.745del (p.His249fs)	HEAT 3	FS, unknown inheritance	Unpublished	1433639
c.489_498del (p.His163fs)	HEAT 1-2	FS, unknown inheritance	Unpublished	454865
c.363_373del (p.Ala122fs)	HEAT 1-2	FS, unknown inheritance	Unpublished	454864
c.8C>T (p.Ala3Val)	N-terminus	Missense, homozygous	Unpublished	
c.208C>T (p.Gln70Ter)	C-terminus	nonsense, unknown inheritance	Unpublished	843828
c.55dup (p.Ala19fs)	N-terminus	FS, compound heterozygous	PMID:29363216†	689528

*Location, HEAT-repeat motif site of the variant

†Reported as compound heterozygous (p.Ala1fs/Cys500Phe)

‡Reported as compound heterozygous (Met703FS/ Trp577Cys)

Abbreviations, FS, frame shift; Ter, termination