

Table SI. AAGAB mutations identified in the current study

	Nucleotide change	Protein change	Computational analysis	Exon	Protein position	Family (patient)	References
Affecting start codon	c.1A>G c.2T>A	p.0? p.0?	Probably damaging deleterious Probably damaging deleterious	1 1	N-terminal N-terminal	PPKP 10 (66651), PPKP 4 (68077, 71005) PPKP 3 (66350, 66351)	Current study Current study
Missense	c.1-1_12del c.415G>A	p.0? p.Val139Ile	Probably damaging deleterious Probably damaging tolerated	-1/1 4	N-terminal Between Rab-like GTPase and adaptin-binding domain	PPKP 2 (66352) PPKP 6 (69352, 71033, 71034, 71035, 76427)	Current study rs138601241 MAF: A=0.001/2
Frameshift	c.77delT c.506_507insAA c.17_73+17delCCT(68bp)GCC	p.Ile26Thrfs*11 p.Asn169Lysis*7 p.?		2 5 1/+1	N-terminal Adaptin-binding N-terminal	PPKP 1 (65325, 65604, 66550, 72382) PPKP 12 (76331) PPKP 14 (78323)	Current study Current study Current study
Nonsense	c.512G>A c.370C>T c.481C>T	p.Trp171* p.Arg124* p.Arg161*	Exon skipping	5 4 5	Adaptin-binding Rab-like GTPase Adaptin-binding	PPKP 11 (73417) PPKP 9 (65536, 72608) PPKP 5 (69783, 70445)	Current study Giehl et al. 2012 (1) Giehl et al. 2012 (1)
Splice-site	c.73+1G>T c.451+1G>A	p.? p.?	Exon skipping Exon skipping	+1 +4	N-terminal Between Rab-like GTPase and adaptin-binding domain	PPKP 13 (76330) PPKP 8 (70562, 73578) PPKP 7 (69354)	Pohler et al. 2012 (12) Current study Eytan et al. 2014

PPKP: punctate palmoplantar keratoderma.