

Supplementary Table 2: Characteristics of UNC45A variants

	Variant NM_018671.4	Consequence	SIFT	POLYPHEN	Mutation taster	CADD	gnomAD Allele frequency
P1							
P5	c.710T>C	Leu237Pro	Deleterious	Possibly damaging	Disease causing	27.2	3.99e-6
P6							
	c.721C>T	Arg241*	Not scored	Not scored	Not scored	23	Not observed
P2							
	c.2182G>A	Glu728Lys	Deleterious	Probably damaging	Disease causing	27.4	6.39e-6
	c.1452delinsGCA	Asp484Glufs*17	Not scored	Not scored	Not scored	23	Not observed
P3							
	c.2512G>C	Ala838Pro	Tolerated	Probably damaging	Disease causing	25.5	Not observed
P4	c.689C>G	Thr230Arg	Deleterious	Possibly damaging	Disease causing	25	Not observed

CADD: Combined Annotation Dependent Depletion.