

Supplementary Table SI. Other rare variations in the proband

Number	Gene	Exon	Nucleotide/amino acid changes	Chromosome location
1	NBAS	NM_015909 exon51	c.6712-1G>T -	chr2:15319241
2	NBAS	NM_015909 exon26	c.3071G>A p.G1024D	chr2:15542292
3	GJB	NM_004004 exon2	c.109G>A p.V37I	chr13:20763612
4	VWF	NM_000552 exon16	c.2024C>T p.P675L	chr12:6161871
5	MAGI2	NM_012301 exon12	c.2117C>T p.T706M	chr7:77824343
6	PIEZO1	NM_001142864 exon45	c.6646C>G p.L2216V	chr16:88783445