

	<i>DIAPH2</i> variant
Predictor	p.I290V
SIFT	Tolerated
Polyphen 2 HumDiv	Possibly Damaging
Polyphen 2 HumVar	Possibly Damaging
MutationTaster	Disease causing
Mutation Assessor	FI M
FATHMM	Damaging
Condel	Deleterious
CADD	23.4

**Figure S11. Pathogenicity prediction of the p.I290V missense variant with 8 commonly used software.** FI, Functional Impact of a variant; CADD, Combined Annotation Dependent Depletion. FI is ranked as: high (H), medium (M) for predicted functional variants and low (L) for predicted non-functional variants. CADD Scaled C-score represents the PHRED-like  $[-10 \cdot \log_{10}(\text{rank}/\text{total})]$  score, ranking a variant relative to all possible substitutions of the human genome ( $8.6 \times 10^9$ ).