

Suppl. Table II. *NLRP12* variant characteristics

Genomic position ^a	Chr19:54314056
Coding sequence alteration ^b	c.857C>T
Amino acid alteration	p.Pro286Leu
Allele count in the gnomAD database	40
Allele frequency in the gnomAD database	1.42x10 ⁻⁴
SIFT ^c	0
LRT ^d	0.015882
GERP++ ^e	4.47
PhyloP ^f	0.944
Polyphen ^g	0.877
CADD ^h	23.1

Predictive analyses were performed with GATK analysis toolkit (Ref PMID: 20644199) and CADD (Ref PMID: 30371827)

^a Positions refer to Hg19 (GRCh37).

^b Positions refer to GenBank transcript NM_144687.4.

^c Function prediction tool based on protein sequence conservation among homologs. Variants with scores between 0 and 0.05 are considered deleterious.

^d DNA sequence evolutionary model expressed as a p-value.

^e GERP++NR score: DNA conservation score.

^f Vertebrate PhyloP scores. Values vary between -20 and +9.873. Sites predicted to be conserved are assigned positive scores, while sites predicted to be fast-evolving are assigned negative scores.

^g Polyphen2_HDIV_score: variants with scores between 0.85 and 1.0 are predicted to be damaging with high confidence.

^h CADDv1.3 scores ranges from 1 to 99, with a higher score indicating greater deleteriousness.