

Supplement Table 1. Allele frequencies and bioinformatics analysis of the *IFIH1* variants identified in this study

Case No	<i>IFIH1</i> variants (NM_022168.4)	MAF				D:A ^a	PP2_Var	<i>In silico</i> missense prediction						ACMG (scoring)
		gnomAD v4.1.0	gnomAD v2.1.1	gnomAD EAS v2.1.1	gnomAD v2.1.1 controls			Mutation Taster	CADD	REVEL	FATHMM_MKL	Geno Canyon	GERP	
1	c.2906G>T/p.Gly969Val	-	-	-	-	21:23	PD (1)	DC (1)	D (33)	D (0.897)	D (0.991)	D (1)	C (4.92)	LP (PS2+PM2+PP3)
2	c.2966T>A/p.Val989Glu	3.424×10 ⁻⁶	-	-	-	19:23	PD (1)	DC (1)	D (28.8)	D (0.605)	D (0.992)	D (1)	C (5.45)	LP (PS2+PM2+PP3)
3	c.263G>A/p.Ser88Asn	4.788×10 ⁻⁶	1.591×10 ⁻⁵	2.175×10 ⁻⁴	1.828×10 ⁻⁵	2:23	B (0)	P (1)	T (0.004)	T (0.114)	T (0.035)	D (1)	NC (-5.69)	US (-)
	c.1046A>G/p.Lys349Arg	2.557×10 ⁻³	2.899×10 ⁻³	1.506×10 ⁻⁴	3.179×10 ⁻³	11:23	B (0.345)	DC (0.862)	T (15.54)	T (0.039)	D (0.875)	D (1)	C (5.96)	US (PP3)
4	c.1709T>G/p.Met570Arg	7.026×10 ⁻⁷	-	-	-	4:23	B (0.218)	DC (0.843)	T (14.57)	T (0.051)	D (0.823)	T (0.979)	C (3.24)	US (PM2+PP3)
	c.2366C>T/p.Thr789Ile	2.236×10 ⁻⁵	1.492×10 ⁻⁴	2.108×10 ⁻³	1.752×10 ⁻⁴	19:23	PD (1)	DC (1)	D (29.7)	D (0.570)	D (0.996)	D (1)	C (5.72)	US (PP3)
5	c.2069T>C/p.Leu690Pro	1.869×10 ⁻⁵	1.030×10 ⁻⁴	1.390×10 ⁻³	1.126×10 ⁻⁴	18:23	PD (1)	DC (1)	D (25.1)	D (0.482)	D (0.985)	D (1)	C (5.36)	US (PP3)
	c.2232T>A/p.Phe744Leu	9.355×10 ⁻⁵	2.547×10 ⁻⁴	3.358×10 ⁻³	2.827×10 ⁻⁴	13:23	PD (0.984)	DC (0.999)	D (22.8)	T (0.272)	D (0.936)	T (0.666)	C (3.19)	US (PP3)

Abbreviations: ACMG, American College of Medical Genetics and Genomics; B, benign; C, conserved; CADD, combined annotation-dependent depletion; D, damaging; DC, disease_causing; EAS, East Asian; GERP, genomic evolutionary rate profiling; gnomAD, the Genome Aggregation Database; LP, likely pathogenic; MAF, minor allele frequency; NC, non-conserved; P, polymorphism; PD, probably_damaging; PM2, Absent in population databases; PP2_Var, polyphen2_HVAR; PP3, Multiple lines of computational evidence support a deleterious effect on the gene/gene product; PS2, De novo in a patient with the disease and no family history; REVEL, Rare Exome Variant Ensemble Learner; T, tolerable; US, uncertain significance.

^aNumber of algorithms predicted to be deleterious: total *in silico* algorithms, which was retrieved from the website <http://www.genemed.tech/varcards/welcome/index>. Due to space limitations, only seven typical results were indicated in this table.