Table S3. Pathogenic variants in *TSPAN12* gene

Nucleotide change	Amino acid change	N. of families identified in this study	Minor allele frequency			*In-silico* prediction						Conserved domain	Associated phenotype	Known variant [reported in our earlier study]	Pathogenicity (evidenced criteria)*
			gnomAD [27]	HGVD [28]	TOMMO 3.5kJPN v2 [29]	GERP++ [33]	SIFT [34]	M-CAP [35]	REVEL [36]	Polyphen 2 HumDIV [37]	CADD [38]				
c.194C>T	p.P65L	1	NL	NL	NL	5.99	0.02	0.053	0.387	0.386	22.9	(CDD: 189507)	FEVR(DI)	Yes	Likely pathogenic (PM1+PM2+PP2+PP3)
c.232G>A	p.G78R	1	NL	NL	NL	5.99	0	0.260	0.710	0.996	29.8	(CDD: 189507)	FEVR(AD)	Yes	Pathogenic (PS1+PM1+PM2+PP1+PP2+PP3)
c.282A>G	p.A94=	1	NL	NL	NL	NL	NA	NA	NA	NA	15.5	NA	FEVR(DI)	No	Likely pathogenic (PVS1+PM2)
c.338G>A	p.W113*	1	NL	NL	NL	5.25	NA	NA	NA	NA	50.0	NA	FEVR(SP)	No	Likely pathogenic (PVS1+PM2)
c.380_385dup	p.D127_M128dup	1	NL	NL	NL	NL	NA	NA	NA	NA	NA	NA	FEVR(AD)	No	Likely pathogenic (PM1+PM2+PM4+PP1)
c.402G>C	p.R134S	1	NL	NL	NL	2.34	0	0.039	0.404	0.014	22.4	(CDD: 48421)	FEVR(AD)	No	Likely pathogenic (PM1+PM2+PP1+PP2+PP3)
c.419T>A	p.L140*	6	NL	NL	NL	5.89	NA	NA	NA	NA	37.0	NA	FEVR(AD/SP)	[24]	Pathogenic (PVS1+PM2+PP1)
c.644delG	p.R215Kfs*9	1	NL	NL	NL	NL	NA	NA	NA	NA	-	NA	FEVR(AD)	No	Pathogenic (PVS1+PM1+PM2+PP1)
c.734T>C	p.L245P	1	0.0000	NL	NL	5.68	0	0.066	0.545	0.997	28.4	adjacent to	FEVR(AD)	[24]	Likely pathogenic

												(CDD: 189507)			(PM1+PM2+PP1+PP2+PP3)
c.738G>A	p.W246*	1	NL	NL	NL	5.68	NA	NA	NA	NA	40.0	NA	FEVR(AD)	Yes	Pathogenic (PVS1+PS1+PM2+PP1)

Underlined values are classified as "not deleterious" according to the cutoff values ([33]-[37]), AD, autosomal dominant; CDD, Conserved Domain Database (http://www.ncbi.nlm.nih.gov/RefSeq/, [31]); DI, digenic inheritance; FEVR, familial exudative vitreoretinopathy; NA, not applicable; NL, not listed; SP, sporadic.

* Based on the recommendation of the American College of Medical Genetics and Genomics (ACMG) standard and guidelines [32].