

Table S14. Genetic and clinical characteristics of the 108 variant-positive probands with FEVR (AD- and AR- variants in *LRP5* gene separated)

	<i>FZD4</i>	<i>AD-LRP5</i>	<i>AR-LRP5</i>	<i>TSPAN12</i>	<i>NDP</i>	Di-genic
	n = 39 (36.1%)	n = 25 (23.1%)	n = 13 (27.8%)	n = 13 (27.8%)	n = 13 (27.8%)	n = 5 (4.6%)
Male	20 (51.3%)	13 (52.0%)	7 (53.8%)	9 (69.2%)	13 (100.0%)	4 (80.0%)
Female	19 (48.7%)	12 (48.0%)	6 (46.2%)	4 (30.8%)	0 (0.0%)	1 (20.0%)
Familial	31 (79.5%)	15 (60.0%)	5 (38.5%)	11 (84.6%)	5 (38.5%)	2 (40.0%)
Sporadic	8 (20.5%)	10 (40.0%)	8 (61.5%)	2 (15.4%)	8 (61.5%)	3 (60.0%)
Infantile case	29 (74.4%)	16 (64.0%)	11 (84.6%)	9 (69.2%)	11 (84.6%)	5 (100.0%)
Juvenile or adult case	10 (25.6%)	9 (36.0%)	2 (15.4%)	4 (30.8%)	2 (15.4%)	0 (0.0%)
Syndromic	1 (2.6%)	1 (4.0%)	1 (7.7%)	0 (0.0%)	8 (61.5%)	0 (0.0%)
Non-syndromic	38 (97.4%)	24 (96.0%)	12 (92.3%)	13 (100%)	5 (38.5%)	5 (100.0%)
Symmetry*	19 (48.7%)	12 (48.0%)	7 (53.8%)	6 (46.2%)	10 (76.9%)	0 (0.0%)
Asymmetry*	20 (51.3%)	13 (52.0%)	6 (46.6%)	7 (53.8%)	3 (23.1%)	5 (100.0%)

Stage of more severe eye						
Stage 1	2 (5.1%)	2 (8.0%)	0 (0%)	2 (15.4%)	0 (0%)	0 (0%)
Stage 2	2 (5.1%)	1 (4.0%)	0 (0%)	0 (0%)	0 (0%)	0 (0%)
Stage 3	9 (23.1%)	4 (16.0%)	3 (23.1%)	2 (15.4%)	3 (23.1%)	0 (0%)
Stage 4	18 (46.2%)	10 (40.0%)	6 (46.2%)	5 (38.5%)	2 (15.4%)	3 (60.0%)
Stage 5	4 (10.3%)	5 (20.0%)	4 (30.8%)	2 (15.4%)	8 (61.5%)	2 (40.0%)
Stage R	4 (10.3)	3 (12.0%)	0 (0%)	2 (15.4%)	0 (0%)	0 (0%)

AD, autosomal dominant; AR autosomal recessive; R, rhegmatogenous retinal detachment. *R was assigned to the original stage.