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| Supplemental table 1. Silico analysis of novel variants identified in patients with FEVR | | | | | | | | | | | |
| Pt | Sex | Gene name | Variants (Protein) | Effect | Inheritance/allele status | MAF | SIFT | PolyPhen\_2 | MutationTaster | GERP++ | Cosegregation |
| 3 | M | NDP | c.17T> C(p.L6P) | Missense | XL/hemi | NA | Damaging | Probably damaging | Disease causing | Conserved | Mother |
| 9 | M | LRP5 | c.3892T>C (p.C1298R) | Missense | AD/het | NA | Damaging | Probably damaging | Disease causing | Conserved | Father |
| 13 | M | LRP5 | c.4178dup T(p.I1394H fs\*156) | Frameshift | AD/het | NA | - | - | - | - | Father |
| 14 | M | TSPAN12 | c.300G> C(p.L100F) | Missense | AD/het | NA | Damaging | Probably damaging | Disease causing | Nonconserved | Father |
| 17 | M | FZD4 | c.347C>T (p.P116L) | Missense | AD/het | NA | Damaging | Probably damaging | Disease causing | Conserved | Mother |
| 19 | M | LRP5 | c.1676A>G(p.Y559C) | Missense | AR/het | NA | Damaging | Probably damaging | Disease causing | Conserved | Mother |
| AD, autosomal dominant; AR, autosomal recessive; FEVR, familial exudative vitreoretinopathy; MAF, minor allele frequency; XR, X-lined recessive. | | | | | | | | | | | |