**S2 Table.** Summary of novel exome variants that co-segregated with disease in the family trio (Fig. 1)

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| Genomic location  (hg19 assembly) | Gene symbol | cDNA variant  in I:1 and II:4 | Protein variant | SIFT | PolyPhen-2 | Glaucoma locus | Co-segregation in pedigree (recombinant) |
| 1:90,050,085 | *LRRC8B* | c.1876A>G | p.Ile626Val | Not tolerated | Probably damaging |  | No (II:5, II:7) |
| 1:155,874,190 | *RIT1* | c.392A>T | p.Phe133Tyr | Tolerated | Benign |  | No (II:5) |
| 1:201,190,860 | *IGFN1* | c.10181C>T | p.Ala3394Val | Tolerated | Possibly damaging |  | No (II:7, II:9) |
| 2:56,144,899 | *EFEMP1* | c.418C>T | p.Arg140Trp | Not tolerated | Probably damaging | GLC1H | Yes |
| 2:152,298,449 | *RIF1* | c.1678C>A | p.Pro560Thr | Not tolerated | Probably damaging |  | No (II:1, II:7) |
| 3:49,200,677 | *CCDC71* | c.965G>A | p.Ala322Val | Tolerated | Benign | GLC1L | No (II:1, II:5, II:10) |
| 5:90,025,579 | *GPR98* | c.10547T>C | p.Ile3516Thr | Tolerated | Benign |  | No (II:5, II:10) |
| 6:31,084,304 | *CDSN* | c.1088G>A | p.Ala363Val | Damaging | Benign |  | No (II:1, II:3, II:9) |
| 11:66,083,293 | *CD248* | c.1205C>G | p.Gln402His | Tolerated | Benign |  | Yes |
| 19:9,066,693 | *MUC16* | c.20753T>C | p.Asp6918Gly | Damaging | Benign |  | No (II:5, II:7, II:10) |
| 19:44,223,572 | *IRGC* | c.862G>A | p.Ala288Thr | Tolerated | Benign |  | No (II:5, II:7, II:10) |
| 19:51,218,952 | *SHANK1* | c.495C>G | p.Asn165Lys | Not tolerated | Probably damaging |  | No (II:5, II:7, II:10) |
| 22:46,751,439 | *TRMU* | c. 970C>G | p.Asp324Glu | Tolerated | Benign |  | No (II:3, II:5) |