**Suppl Table 1: Details of the exonic variants shared by the two sisters with GAPO and Congenital glaucoma**

|  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Pedigree** | **Variant** | **rs number** | **gene** | **DNA change** | **Amino acid**  **change** | **SIFT Score** | **Polyphen2**  **score** | **CADD**  **score** | **ACMG**  **Classification#** | **Clinical significance** |
| Case 1 and Case 2 | 22:21567413-G/A | [rs1339860092](http://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=rs1339860092) | *GGT2* | NM\_001351304.1:c.976C>T | p.Arg326Trp | 0.001 |  |  | PM2 | US |
| 5:140502997-A/C | rs782542981 | *PCDHB4* | NM\_018938.4:c.1417A>C | p.Ser473Arg | 0.12 | 0.66 | 16.52 | BS1,BP1,BP4 | likely benign |
| 2-120714402-TTT- | [rs76723123](http://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=rs76723123) | *PTPN4* | NM\_002830.4:c.1981-5\_1981-3del |  |  |  | 1.57 | BS1 | likely benign |
| 5:135388650- C/T | [rs201210696](http://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=rs201210696) | *TGFBI* | NM\_000358.3:c.968C>T | p.Ala323Val | 0.027 | 0.47 | 24.1 | PP2, PP3, BS2 | US |
| Case 1 , Case 2 and Mother | 1:6512085 -G/A | [rs781386845](http://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=rs781386845) | *ESPN* | NM\_031475.3:c.2254G>A | p.Glu731Lys | 0.0 | 0.99 | 32 | PM2, PP3 | US |
| 1:5927818 -G/A | [rs560329867](http://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=rs560329867) | *NPHP4* | NM\_001291593.2:c.1915C>T | p.Pro639Ser | 0 | 0.5 | 25.8 | PM2, PP3, BP6 | US |
| 12:104123924 -A/G | [rs200802359](http://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=rs200802359) | *STAB2* | NM\_017564.10:c.5113A>G | p.Ile1705Val | 0.72 | 0.028 | 10.25 | BS1,BP1,BP4 | likely benign |
| 15:78882937 | [rs745841148](http://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=rs745841148) | *CHRNA5* | NM\_000745.3:c.1208\_1212del | p.Ile403fs | - | - | 32 | PVS1  PP3 | US |
| 5:149509395-G/A | [rs142992960](http://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=rs142992960) | *PDGFRB* | NM\_001355017.1:c.1021C>T | p.Arg502Trp | 0.004 | 0.39 | 26.7 | BS1, BS2, BP6 | benign |
| 1:16461688-T/C splice\_region\_variant | [rs374403114](http://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=rs374403114) | *EPHA2* | NM\_001329090.2:c.1267-4A>G | Splice variant |  |  | 0.22 | BS1, BS2 | benign |
| 16:56377830-G/A | [rs201789251](http://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=rs201789251) | *GNAO1* | NM\_138736.3:c.1033G>A | p.Ala345Thr | 0.04 |  |  | PM1, PP2, PP3  BS1,BS2 | benign |
| 12:119624853 -G/A | [rs754993844](http://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=rs754993844) | *HSPB8* | NM\_014365.3:c.391G>A | .Glu131Lys | 0.003 | 0.988 | 32 | BS1, BS2, PP3 | benign |
| 5:148421169 AGA/- | [rs763476083](http://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=rs763476083) | *SH3TC2* | NM\_024577.4:c.536TCT[1] | Phe180del | - | - | 17.9 | PM2,PM4, PP3 | likely pathogenic |
| 8:106646469 -T/ C | [rs552410913](http://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=rs552410913) | *ZFPM2* | NM\_012082.4:c.421-5T>C | 421-5T>C | - | - | 13.35 | BS2 | benign |
| 15:84324473 -G/C |  | *ADAMTSL3* | NM\_207517.3):c.-33-8G>C | c.-33-8G>C | - | - |  | BP4 | US |
| Case 1 , Case 2 and Father | 6:111702554   -T/G | [rs3218579](http://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=rs3218579) | *REV3L* | NM\_002912.5:c.1190A>C | p.Gln397Pro | 0.01 | 0.96 | 26.3 | BS1, BP1, BP6 | likely benign |
| 7:6045616  -G/T |  | *PMS2* | NM\_000535.7:c.70C>A | p.His24Asn | 0.009 | 0.98 |  | PM2, PP3 | likely pathogenic |
| 10:74928067   initiator\_codon\_variant |  | *FAM149B1* |  |  |  |  |  |  |  |
| 19:55671306- C/T | [rs549436949](http://www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=rs549436949) | *DNAAF3* | NM\_001256714.1:c.1325G>A | p.Cys422Tyr | 0.066 | - | - | PM2, BP4 | US |

SIFT- Sorting Intolerant from Tolerant, CADD- Combined Annotation Dependent Depletion, Polyphen2- Polymorphism Phenotyping v2, **# -**Richards et al 2015, US-Uncertain significance