Supplemental Table 1. Summary of heterozygous *CYP1B1* variants of uncertain significance and patient features.

***CYP1B1* variant**[a]	**Race/ ethnicity**	**Functional prediction**[b]	**Allele frequency**[c]	**# homozygotes in ExAC**[d]	**Patient phenotype**	**Co-segregation**	**Previously reported**[e]
c.1462G>C, p.(Ala488Pro)	Caucasian	Damaging by 3/5 (S, PP, MA)	Not present	Not present	Isolated corectopia	Also present in unaffected mother	No
c.241T>A, p.(Tyr81Asn)	Caucasian	Damaging by 4/5 (S, PP, MT, MA)	159/22812	1 Eu, 2 SA, 1 Fn	ASD, congenital glaucoma,	Also present in unaffected mother	Yes- POAG (Het) and PCG (Het, CHet) (1-8)
c.1328C>G, p.(Arg443Gly)	African American	Benign by 5/5	86/66736	14 Af	Unilateral Peters anomaly with microphthalmia,	Also present in unaffected brother	Yes- POAG (Het), PCG (Het), and controls (Het) (2-5, 9-12)

ASD=Anterior segment dysgenesis; PCG=Primary congenital glaucoma; POAG=Primary open angle glaucoma; novel variants are shown in **bold** font

[a] Nucleotide numbering is relative to reference sequence NM_000104.3 where +1 is the A of the ATG initiation codon
[b] Five prediction algorithms (SIFT (S), PolyPhen2 (PP), MutationTaster (MT), MutationAssessor (MA), FATHMM (F)) from dbNSFP 2.9 were accessed through SNP & Variation Suite (Golden Helix, Bozeman, MT)
[c] Allele frequency for Caucasian populations in ExAC (Exome Aggregation Consortium; http://exac.broadinstitute.org/)
[d] Number of homozygotes present in ExAC is noted; Af: African, Eu: European, SA: South Asian, Fn: Finnish
[e] Phenotype and zygosity in previously reported individuals; Het=heterozygous change; CHet=compound heterozygous change

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