Supplementary material for Tran et al., "Primary Congenital Glaucoma in Vietnam: Analysis and identification of novel CYP1B1 variants", *Ophthalmic Genetics*, 2019.

Patient No.	Nucleotide change	Amino acid change	Mutation type	Heterozygous/Homozygous
G02		Whole CYP1B1 deletion		Homozygous
G56		Whole CYP1B1 deletion		Homozygous
G10	c.475C>T	Q159X	Nonsense	Heterozygous
	c.652G>C	D218H	Missense	Heterozygous
G09	c.475C>T	Q159X	Nonsense	Heterozygous
	c.490C>T	Q164X	Nonsense	Heterozygous
	c.652G>C	D218H	Missense	Heterozygous
G11	c.256C>A	Q86K	Missense	Heterozygous
	c.475C>T	Q159X	Nonsense	Heterozygous
G15	c.571DelC	L191Sfs*4	Frameshift	Homozygous
G20	c.80T>A	L27Q	Missense	Heterozygous
	c.182G>A	G61E	Missense	Heterozygous
G21	c.256C>A	Q86K	Missense	Heterozygous
	c.592G>A	V198I	Missense	Heterozygous
G24	c.685G>A	E229K	Missense	Heterozygous
	c.724G>A	D242N	Missense	Homozygous
G40	c.652G>C	D218H	Missense	Heterozygous
	c.685G>A	E229K	Missense	Heterozygous
G08	c.256C>A	Q86K	Missense	Heterozygous
G19	c.397G>A	A133T	Missense	Heterozygous
G43	c.256C>A	Q86K	Missense	Heterozygous
G44	c.1094G>A	G365E	Missense	Heterozygous

Ten patients (11.76%) were found to have two likely pathogenic mutations causative of PCG (G02, G56, G10, G09, G11, G15, G20, G21, G24, G40) while the other four (G08, G19, G43, G44) have only one mutation in heterozygous state.

Patient G09 has three different most likely disease-causing variants Q159X, Q164X, D218H, in which parental testing need to be carried out to determine the cis/tran state of these mutations.

Patient G24 carries the variant p.D242N in homozygous and the p.E229K change in heterozygous state. However, E229K mutation were reported to be likely benign, that would leave D242N to be likely causative of PCG in this patient.