

Table S1. Criteria used to classify sequence variants according to ACMG guidelines (46).

Variant ID	Evidences of pathogenicity	Class
M1	1 moderate evidence (PM2) and 4 supporting evidences (PP1-PP2-PP3-PP4)	Likely pathogenic
M2	1 moderate evidence (PM2) and 4 supporting evidences (PP1-PP2-PP3-PP4)	Likely pathogenic
M3	1 very strong evidence (PVS1), 1 moderate evidence (PM2) and 3 supporting evidences (PP1-PP3-PP4)	Pathogenic
M4	2 moderate evidences (PM2-PM3) and 3 supporting evidences (PP1-PP2-PP4)	Likely pathogenic
M5	4 supporting evidences (PP1-PP2-PP3-PP4)	Uncertain significance
M6	1 moderate evidence (PM2) and 4 supporting evidences (PP1-PP2-PP3-PP4)	Likely pathogenic

M: mutation.

Table S2: Incidental findings detected by NGS in inherited retinal disorders genes other than Bradet Biedl and Usher syndromes.

Index Patient (corresponding disease)	Gene	Associated IRD	Status	Exon	rs ID	Nucleotide Exchange	Amino Acid Change	Frequencies	PolyPhen-2	SIFT
FA4: V.3 (BBS)	<i>CDH23</i>	USH	HTZ	67	rs76399310	c.9587_9589delACA	p.Asn3197del	0.0000643 (gnomAD)	-	-
	NM_022124.5				0			0.0000159 (TOPMed)		
								Never Hom		
	<i>IFT140</i>	RP or LCA	HTZ	20	rs20087669	c.2569G>A	p.Gly857Ser	0.003545 (gnomAD)/8HOM	Probably damaging	D
	NM_014714.4				6			0.001911 (TOPMed)/1HOM		
	<i>RBP3</i>	RP	HTZ	1	rs14428991	c.1795A>G	p.Ile599Val	0.001144 (gnomAD)	Benign	D
	NM_002900.3				2			0.0009795 (TOPMed)		
								Never Hom		
	<i>CDHR1</i>	CRD	HTZ	8	rs74738807	c.764T>A	p.Val255Glu	0.00002386 (gnomAD)	Possibly damaging	D
	NM_033100.4				6			0 (TOPMed)		

Never Hom

FB22: II.1	<i>HGSNAT</i>	RP	HTZ	1	-	c.11C>A		p.Ala4Glu	0 (gnomAD)	Benign	-
(BBS)	NM_152419.3								0 (TOPMed)		
									Never Hom		
	<i>EYS</i>	RP	HTZ	4	-	c.476G>C	p.Cys159Ser	p.Cys159Ser	0 (gnomAD)	Probably	D
	NM_001292009.1								0.00000796 (TOPMed)	damaging	
									Never Hom		
	<i>CDH23</i>	USH	HTZ	68	rs200124827	c.9928C>T		p.Arg3310Cys	0.0002158 (gnomAD)	Possibly	D (low
	NM_022124.5								0.0001832 (TOPMed)	damaging	confidence)
FD10: III.3	<i>GUCY2D</i>	LCA	HTZ	4	rs140638938	c.1315G>A		p.Gly439Arg	0.000334 (gnomAD)//1Hom	Probably	D
(USH)	NM_000180.4								0.0000079 (TOPMed)	damaging	
	<i>GUCY2D</i>	LCA	HTZ	20	rs552184470	c.124_129delCTGCTT		p.Leu44_Leu45del	0.001913 (gnomAD)	-	-
	NM_000180.4								/3Hom		

							0.002087 (TOPMed) /3Hom		
<i>PDE6A</i> NM_000440.3	RP	HTZ	14	rs76500719 6	c.1730C>T	p.Thr577Met	0.000007962 (gnomAD)	Possibly damagin g	D
							0.0000079 (TOPMed)		
							Never Hom		
<i>RGS9</i> NM_003835.4	recessive delayed cone adaptatio n	HTZ	5	rs20199788 8	c.314C>G	p.Thr105Arg	0.001901 (gnomAD)//1Ho m	Probably damagin g	D
							0.001776 (TOPMed)		

HTZ: heterozygous; HOM: homozygous; D: deleterious, USH: Usher syndrome; RP: retinitis pigmentosa; LCA: Leber's congenital amaurosis; CRD: cone rod dystrophy.