Table S3: Variants of unsolved probands

Patient ID	Diagnosis	Gene Transcript	Inheritance Pattern	Exon / Intron	Genomic Location[a] and cDNA Change	Amino Acid Change	dbSNP ID or Clinvar Accession Number	Zygosity	Revel Score	ClinVar	ACMG	Reference
P5	Retinitis Pigmentosa	*CEP78* NM_001330691.3	AR	E11	chr9:80869869 c.1369G>T	p.Glu457Ter	rs996333495	Het	-	P	LP PVS1, PM2	-
P8	Usher Syndrome	*USH2A* NM_206933.4	AR	E13:E4	c.[2299del; c.682A>T] (pat) chr1:216420436: chr1:216538397	p.[(Glu767SerfsTer21); (Asn228Tyr)]	rs80338903;-	Het	-;0.3	P;-	LP PVS1,PM2; VUS PM2	PMID: 25375654;-
P14	Pattern Dystrophy	*CFH* NM_000186.4	AD	E22	c.3607C>T chr1:196716354	p.Arg1203Trp	rs145347741	Het	0.12	VUS	VUS PM2, PM1, BP4	PMID: 20106822 (HUS)
P16	Cone Dystrophy	*TRPM3* NM_001366145.2	AD	E26	c.4763T>C chr9:73151266	p.Val1588Ala	rs765049568	Het	0.08	-	VUS PM2, BP4	-
		RP1 NM_1375654.1	AD, AR	E29	c.3980G>A chr8:55778404	p.Rrp1327Ter	rs141992026	Het	-	-	VUS PVS1, BS2	-
P25	Retinitis Pigmentosa	*CDH23* NM_022124.6	AR, DR	E60	c.8624T>C chr10:73567666	p.Ile2875Thr	rs776064526	Het	0.14	VUS	VUS PM2, BP4	-
		ABCA4 NM_000350.3	AR	E42	c.5882G>A chr1:94473807	p.Gly1961Glu	rs1800553	Het	0.76	P/LP	LP PM5, PP2, PP3, PS4	PMID: 34874912
P28	Retinitis Pigmentosa	*ABCA4* NM_000350.3	AR	E7	c.806A>G chr1:94548960	p.Asn269Ser	rs1338260475	Het	0.36	VUS	VUS PM2, PP2	-
P33	Retinitis Pigmentosa	*CDH23* NM_022124.6	AD, DR	E25	c.2926A>G chr10:73464860	p.Ser976Gly	rs372401651	Het	0.4	VUS	VUS PM2	-
				E55	c.7823G>A chr10:73563128	p.Arg2608His	rs202052174	Het	0.44	Conflict	VUS BS1	PMID: 19683999
		PCARE NM_001029883.3	AR	E1	c.3032_3046dup chr2:29294081	p.Ser1015 _Tyr1016insCysLeuPro SerSer	rs1293436955	Het	-	VUS	VUS PM2, PM4	-
				E1	c.3581C>G chr2:29293547	p.Ala1194Gly	rs372953965	Het	0.05	VUS	VUS PM2, BP4	-
P34	Retinitis Pigmentosa	*USP45* NM_001346022.3	AR	E9	c.931_932delAA chr6:99924019	p.Lys311fs	SCV006082520	Het	-	-	LP PVS1, PM2	Novel
		USH2A NM206933.4)	AR	E63	c.13339A>G chr1:215847914	p.Met4447Val	rs139474806	Het	0.18	Conflict	LP PM2, PM5, PM1	PMID: 31872526
		ROM1 NM_000327.4)	AD, AR, DD	E1	c.167C>T chr11:62380920	p.Ser56Phe	rs199847029	Het	0.32	VUS	VUS PM2	-
		KIF3B NM_004798.4)	AD	E2	c.246C>G chr20:30897826	p.Asp82Glu	rs556679698	Het	0.37	-	VUS PM2	-

USH2A variants in P8 are presented in cis, with annotations for each variant—including exon number, dbSNP ID, ClinVar, ACMG classification, and reference information—provided separately, using a semicolon.
[a]:Genomic locations have been added based on GRCh37. Abbrevations: AD: autosomal dominant, AR: autosomal recessive, DD: digenic form, DR: digenic recessive, E: exon, Hem: hemizygous, Het: heterozygous, Hom: homozygous, HUS: Hemolytic uremic syndrome, atypical, IVS: intervening sequence, LP: likely pathogenic, NA: not available, P: pathogenic, Pat: paternal, VUS: variant of uncertain significance, XL: X-linked