Supplementary Table 1A: Pedigree information displayed by functional categories of genes, identifying genetic variants that account for or are likely to account for disease in this cohort.

Family ID	Proband Variant ID Nucleotide changes		Siblings		Other family members	Comments
	Paternal Allele	Maternal Allele	Allele 1	Allele 2		
Ciliary transport and trafficking						
***CEP290* (NM_025114.3)**						
0623	c.4625_4626insCATG	c.2991+1655A>G	WT	WT	-	
1015	c.2991+1655A>G	c.3181_3182del	c.3181_3182del	WT	-	-
1212	c.2991+1655A>G	c.1781T>A	WT	WT	-	
1576	c.5587-1G>C	c.3175dup	c.3175dup	WT	-	-
			c.3175dup	WT		
			c.5587-1G>C	WT		
1950	c.2991+1655A>G	c.297+3A>G	c.2991+1655A>G	WT	2nd cousins (maternal)	
	c.2991+1655A>G	c.297+3A>G	-	-		
***LCA5* (NM_001122769.2)**						
1894	c.1144_1147dup	c.1144_1147dup	c.1144_1147dup	c.1144_1147dup	-	**Consanguinity** Clinical diagnosis of fundus albipunctatus (1) and cone-rod dystrophy (1)
***RPGRIP1* (NM_020366.3)**						
0479	c.1219C>T;1763-8C>G	c.exon19del chr14:g.(21798302_21798377)_(21798551_21799045)del (hg19,NC000014.8)	c.1219C>T; 1763-8C>G	c.exon19del		
1642	c.2935C>T	c.1447C>T	c.1447C>T	c.2935C>T	-	+RPGRIP1 c.1639G>T (benign)
***SPATA7* (NM_018418.4)**						
1543	c.763C>T	c.763C>T	c.763C>T	c.763C>T	-	-
			c.763C>T	WT		
			WT	WT		

***TULP1* (NM_003322.3)**						
1620	WT	c.524dup c.524dup	WT	WT	Carrier c.524dup (maternal first cousin once removed)	Maternal isodisomy of chromosome 6; also harbours a variant in *RP1* (c.1267-1656delinsA p.Ala423Asnfs*11)
2175	c.1081C>T	c.999+5G>C	c.999+5G>C	c.1081C>T	-	-
Visual cycle						
***RDH12* (NM_152443.2)**						
1425	c.316C>T^{NT} and c.697G>C^{NT}		DNWTP		None	Biallelism not determined
***RPE65* (NM_000329.2)**						
1404	c.951_956del	c.130C>T	-	-	-	
1723	c.1040G>C^{NT} and c.726-1G>A^{NT}		-	-	Aboriginal family	Biallelism not determined + RPGRIP1 c.1767G>T p.Gln589His
Phototransduction cascade						
***GUCY2D* (NM_000180.3)**						
0535	c.307G>A	c.2595del	-	-	-	
1836	c.2302C>T	c.2516del	-	-	-	-
2019	c.91dup	c.307G>A	-	-	-	-
2123	c.2646C>G	c.2383C>T	-	-		+ (paternal) interstitial deletion 2q13
2599	c.2302C>T	c.2302C>T	-	-		+ GUCY2D c.2345T>A (benign)

Cell – cell interaction						
***CRB1* (NM_201253.2)**						
0306	c.1793del	c.2843G>A	c.1793del c.1793del	c.2843G>A WT	-	-
2274	c.2843G>A	c.613_619del[BD]	c.613_619del	WT	-	Maternal DNA not available; allele identified in sibling
Neuroprotection						
***NMNAT1* (NM_022787.3)**						
1565	c.769G>A	c.507G>A	-	-	-	-
1819	c.500A>G	c.364del	-	-	-	Paternal c.364delA Maltese descent
1965	c.364del	c.769G>A	c.364del	WT	-	Paternal c.364delA Maltese descent
Protein chaperones and trafficking						
***AIPL1* (NM_014336.3)**						
2030	c.356_359del[BD]	c.834G>A	c.356_359del c.356_359del WT	c.834G>A WT WT		Paternal DNA not available; allele identified in sibling
2272	c.834G>A[**BD**]	c.277-2A>G	c.834G>A	c.277-2A>G	Carrier c.834G>A (paternal great aunt)	Paternal DNA not available; allele identified in paternal family

Pedigree ID in Bold indicates simplex case. BD = by default (parental DNA not available but variant detected in related family member); DNWTP = did not wish to participate; NT = not tested; WT = wildtype alelle. All heterozygous and homozygous WT individuals were unaffected.