

Supplementary Table 3. Summary of candidate variants in this cohort

Sample ID	Candidate Gene(s)	Transcript	Nucleotide Change	Amino Acid Change	Genotypes (Sanger confirmed)	HGMD or dbSNP IDs	References
RD13-01	<i>GUCY1A1</i>	NM_000409.3	c.296A>G	p.Y99C	Heterozygous	CM980960	Payne et al. [1998]
RD1-07	<i>PRPF31</i>	NM_015629.3	c.220C>T	p.Q74*	Heterozygous	CM063073	Sullivan et al. [2006]
RD20-06	<i>PRPH2</i>	NM_000322.4	c.514C>T	p.R172W	Heterozygous	CM930639	Poloschek et al. [2010]
RD20-07	<i>PRPH2</i>	NM_000322.4	c.514C>T	p.R172W	Heterozygous	CM930639	Poloschek et al. [2010]
RD2-01	<i>RGR</i>	NM_001012720.1	c.824dupG	p.L276N*77	Heterozygous	C1993291	Morimura et al. 1999
RD11-03	<i>RHO</i>	NM_000539.3	c.574dup	p.Y192Lfs*139	Heterozygous	Novel	
RD14-07	<i>TOPORS</i>	NM_005802.4	c.2474dupA	p.Y825*	Heterozygous	CSI075704	Chakarova et al [2007]
RD10-02	<i>RPIL1</i>	NM_178857.5	c.133C>T	p.R45W	Heterozygous	CM105618	Akahori et al [2010]
RD5-08	<i>CERKL</i>	NM_001030311.2	c.1045_1046del	p.M349Vfs*20	Heterozygous	Novel	
			c.316C>T	p.R106C	Heterozygous	Novel	
RD14-08	<i>USH2A</i>	NM_206933.2	c.2276G>T	p.C759F	Heterozygous	CM001372	Garcia-Garcia et al. [2011]
			c.4108G>C	p.Va370L	Heterozygous	Novel	
RD20-08	<i>USH2A</i>	NM_206933.2	c.2299delG	p.E766Sfs*21	Heterozygous	CD982997	O'Sullivan et al. [2012]
			c.2276G>T	p.C759F	Heterozygous	CM001372	Garcia-Garcia et al. [2011]
RD20-05	<i>CACNA1F</i>	NM_005183.3	c.2576+1G>A	IVS20+1G>A	Hemizygous	Novel	
RD6-08	<i>CHM</i>	NM_000390.2	c.49+2dupT	IVS1+2dupT	Hemizygous	CI137323	Ramsden et al. [2013]
RD12-02	<i>RPGR</i>	NM_000328.2	c.1088_1089delinsA	p.V363Dfs*18	Hemizygous	Novel	
RD4-04	<i>ABCA4</i>	NM_000350.2	c.1804C>T	p.R602W	Homozygous	CM990025	Ribeiro-Alvarez et al. [2013]
RD6-01	<i>BBS1</i>	NM_024649.4	c.1169T>G	p.M390R	Homozygous	CM021489	Baveillette et al. [2003]
RD12-06	<i>CERKL</i>	NM_001030311.2	c.481+2T>G	IVS2+2T>G	Homozygous	CS140556	Glockle et al. [2013]
RD4-06	<i>EYS</i>	NM_001142800.1	c.2259+1G>A	IVS14+1G>A	Heterozygous	CS150721	Bonilha et al. [2014]
			c.6137G>A	p.W2046*	Heterozygous	Novel	
RD6-07	<i>EYS</i>	NM_001142800.1	c.8473_8474insT	p.V2804fs	Heterozygous	Novel	
			c.1153T>G	p.C385G	Heterozygous	Novel	
RD12-05	<i>EYS</i>	NM_001142800.1	c.6416G>A	p.C2139Y	Heterozygous	CM102730	Audo et al. [2010]
			c.7868G>A	p.G2623E	Heterozygous	Novel	
RD13-05	<i>GUCY2D</i>	NM_000180.3	c.2375C>T	p.P792L	Heterozygous	rs763774686	
RD11-08	<i>IMPDH1</i>	NM_000883.3	c.931G>A	p.D311N	Heterozygous	CM020283	Bowne et al. [2002]
RD15-03	<i>IMPDH1</i>	NM_000883.3	c.931G>A	p.D311N	Heterozygous	CM020283	Bowne et al. [2002]
RD5-04	<i>RHO</i>	NM_000539.3	c.68C>A	p.P23H	Heterozygous	CM900197	Dryja et al [1990]
RD13-02	<i>RHO</i>	NM_000539.3	c.68C>A	p.P23H	Heterozygous	CM900197	Dryja et al [1990]
RD12-08	<i>RHO</i>	NM_000539.3	c.561T>G	p.C187W	Heterozygous	Novel	
RD12-07	<i>RHO</i>	NM_000539.3	c.936+1G>T	IVS4+1G>T	Heterozygous	CS920776	Hernan et al. [2011]
RD14-06	<i>PRPH2</i>	NM_000322.4	c.514C>T	p.R172W	Heterozygous	CM930639	Poloschek et al. [2010]
RD12-03	<i>RPE65</i>	NM_000329.2	c.886dup	p.R296Kfs*7	Homozygous	CI107001	Coppieters et al. [2010]
RD12-01	<i>RPGR</i>	NM_000328.2	c.197A>G	p.Q66R	Hemizygous	Novel	
RD6-05	<i>TIMP3</i>	NM_000362.4	c.29T>A	p.L10H	Heterozygous	Novel	
RD20-03	<i>TRPM1</i>	NM_002420.5	c.1197G>A	p.P421=	Heterozygous	CS097758	Audo et al. [2009]
			c.215A>G	p.Y72C	Heterozygous	CM097760	Audo et al. [2009]
RD6-04	<i>TRPM1</i>	NM_002420.5	c.2947_2948delGCinsAT	p.A983I	Heterozygous	Novel	
			c.3125T>G	p.L1042R	Heterozygous	Novel	
RD14-02	<i>USH2A</i>	NM_206933.2	c.11411del	p.P3804Lfs*13	Heterozygous	CD149996	Baux et al. [2014]
			c.8431C>A	p.P2811T	Heterozygous	rs11033529	
RD11-04	<i>USH2A</i>	NM_206933.2	c.13335_13347delinsCTTG	p.E4445_54449delinsDL	Heterozygous	CX104126	McGee et al. [2010]
			c.14402_14403del	p.Y4801Ffs*20	Heterozygous	Novel	
RD14-04	<i>GUCY1A1</i>	NM_000409.3	c.296A>G	p.Y99C	Heterozygous	CM980960	Payne et al. [1998]
RD2-02	<i>RGR</i>	NM_001012720.1	c.824dupG	p.L276N*77	Heterozygous	C1993291	Morimura et al. [2000]
RD10-02	<i>RPIL1</i>	NM_178857.5	c.133C>T	p.R45W	Heterozygous	CM105618	Akahori et al [2010]
RD10-02	<i>RPIL1</i>	NM_178857.5	c.133C>T	p.R45W	Heterozygous	CM105618	Akahori et al [2010]
RD18-02	<i>CERKL</i>	NM_001030311.2	c.1045_1046del	p.M349Vfs*20	Heterozygous	Novel	
			c.316C>T	p.R106C	Heterozygous	Novel	
RD20-02	<i>USH2A</i>	NM_206933.2	c.2299delG	p.E766Sfs*21	Heterozygous	CD982997	O'Sullivan et al. [2012]
			c.2276G>T	p.C759F	Heterozygous	CM001372	Garcia-Garcia et al. [2011]
RD1-12	<i>C2orf71</i>	NM_001029883.2	c.1514G>A	p.W505*	Heterozygous	CM1511740	Yang et al [2015]
			c.3266dup	p.S1090Ifs*17	Heterozygous	Novel	
			c.1375C>T	p.Q459*	Heterozygous	Novel	
			c.1557T>G	p.Y519*	Heterozygous	CM135790	Simeonov et al. [2013]
			c.834C>G	p.Y278*	Heterozygous	CM083852	Hutton et al. [2008]
RD11-05	<i>GUCY1A1</i>	NM_000409.3	c.149C>T	p.P50L	Heterozygous	CM012969	Downes et al. [2001]
RD11-06	<i>GUCY2D</i>	NM_000180.3	c.1724C>T	p.P575L	Heterozygous	CM023932/rs28743021	Small et al. [2008]
			c.1217C>T	p.P406L	Heterozygous	CM910385/rs104894313	Giebel et al. [1991]
			c.1261+1G>A	IVS9+1G>A	Heterozygous	rs140365820	
RD14-05	<i>TRPM1</i>	NM_002420.5	c.1192T>C	p.W398R	Heterozygous	Novel	
			c.3914G>A	p.R1305H	Heterozygous	Novel	
RD14-03	<i>ABCA4</i>	NM_000350.2	c.5714+5G>A	IVS40+5G>A	Heterozygous	CS982057	Creemers et al. [1998]
			c.8600C>T	p.S2867L	Heterozygous	rs145468090	
			c.10552G>A	p.V3518I	Heterozygous	rs75397806	
RD4-05	<i>EYS</i>	NM_001142800.1	c.6138G>A	p.W2046*	Heterozygous	Novel	
RD11-02	<i>EYS</i>	NM_001142800.1	c.5677_5681del	p.Y1893Rfs*12	Heterozygous	Novel	
RD15-01	<i>ABCA4</i>	NM_000350.2	c.4685T>C	p.I1562T	Heterozygous	CM970013	Allikmets et al. [1997]
			c.1753C>T	p.P585S	Heterozygous	CM111852	Fernandez-Martinez et al. [2011]
			c.4574C>T	p.A1525V	Heterozygous	CM078457	Guo & Cooper [2007]
RD14-01	<i>BBS9</i>	NM_198428.2	c.1280C>T	p.A427V	Heterozygous	rs138072724	
			c.1120T>C	p.W374R	Heterozygous	Novel	
			c.5295C>G	p.F1765L	Heterozygous	rs201388114	
RD1-07	<i>ABCA4</i>	NM_000350.2	c.635G>A	p.R212H	Heterozygous	CM020912	Pang et al. [2002]
			c.6529G>A	p.D2177N	Heterozygous	CM970023	Wisniewski et al. [2005]
			c.1552G>A	p.E518K	Heterozygous	CM1410571(DM?)/rs369	Liesz et al. [2015]
RD11-03	<i>CNGA3</i>	NM_001298.2	c.1810C>T	p.Q604*	Heterozygous	Novel	
			c.963T>G	p.N321K	Heterozygous	CM003833/rs149916178	Philp et al. [2009]
RD20-07	<i>CERKL</i>	NM_001030311.2	c.769C>T	p.R257*	Heterozygous	CM040509	Bornancin et al. [2005]
RD14-08	<i>GUCY2D</i>	NM_000180.3	c.2950T>C	p.C984R	Heterozygous	Novel	
RD20-02	<i>CLRN1</i>	NM_174878.2	c.218A>G	p.Q73R	Heterozygous	CM127999	Licastro et al. [2012]
RD12-02	<i>CACNA1F</i>	NM_005183.3	c.1619T>C	p.F540S	Hemizygous	Novel	
			c.1387C>T	p.R463*	Heterozygous	CM110634	Chaki et al. [2011]
RD12-01	<i>RPIL1</i>	NM_178857.5	c.416dup	p.G140Rfs*10	Heterozygous	rs201192645	
			c.721G>A	p.A241T	Heterozygous	CM145799 (DM?)	Fossbakk et al. [2014]
RD13-02	<i>ABCA4</i>	NM_000350.2	c.1610G>A	p.R537H	Heterozygous	CM032805	Jaakson et al [2003]

RD12-07	<i>TYRP1</i>	NM_000550.2	c.1354A>G	p.M452V	Heterozygous	CM081465	Hutton et al. [2008]
RD13-08	<i>ABCA4</i>	NM_000350.2	c.6089G>A	p.R2030Q	Heterozygous	CM990070	Lewis et al. [1999]
	<i>RPGRIPI</i>	NM_020366.3	c.1767G>T	p.Q589H	Heterozygous	CM057749	Fernández-Martínez et al. [2011]
RD15-03	<i>OCA2</i>	NM_000275.2	c.1004C>T	p.T335M	Heterozygous	rs533988694	
RD20-03	<i>OCA2</i>	NM_000275.2	c.1513A>C	p.F505V	Heterozygous	None	UCSC:15-28211959-A-C
RD6-05	<i>TYR_1</i>	NM_000372.4	c.740G>T	p.C247F	Heterozygous	Novel	
RD11-07	<i>OCA2</i>	NM_000275.2	c.1574C>T	p.P525L	Heterozygous	None	UCSC:15-2821898-G-A
RD11-08	<i>TYRP1</i>	NM_000550.2	c.977G>A	p.R326H	Heterozygous	CM135784/rs16929374	
RD13-01	<i>OCA2</i>	NM_000275.2	c.40G>A	p.A14T	Heterozygous	rs368928996	