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Supplemental Data

Whole-Exome Sequencing Identifies *KIZ* as a Ciliary Gene Associated with Autosomal-Recessive Rod-Cone Dystrophy

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SNP	Exon	Nucleotide Exchange	Allele State	Protein Effect	Minor Allele Frequency (n=340 subjects)	Alleles Frequency
rs150243168	1	c.48C>T	heterozygous	p.(=)	T= 0.006	dbSNP: C: 99.862% (2175 / 2178); T: 0.138% (3 / 2179), **EVS** (EA) T=15/C=7753 (AA) T=2/C=3528
rs16982513	4	c.348A>G	5 heterozygous /1 homozygous	p.(=)	G= 0.001	dbSNP: A: 98.320% (2282 / 2321); G: 1.680% (39 / 2321), **EVS** (EA) G=6/A=7834 (AA) G=170/A=3356
rs4815025	5	c.417C>G	148 heterozygous /136 homozygous	p.His139Gln	C=0.38	dbSNP: C: 45.454% (1668.785 / 3671); G: 54.546% (2002.625 / 3671), **EVS** (EA) G=5594/C=2636 (AA) G=1445/C=2365
rs200388739	5	c.505A>G	heterozygous	p.Ser169Gly	G= 0.001	No frequency in dbSNP or 1000Genome, found in Exome chip, NHLBI-ESP , **EVS** (EA) G=1/A=8237
rs75984134	5	c.509T>G	3 heterozygous	p.Met170Arg	G= 0.004	dbSNP: G: 1.437% (33 / 2296); T: 98.563% (2263 / 2296), **EVS** (AA)= G=107/T=3733
rs34305929	5	c.698G>A	heterozygous	p.Gly233Asp	A= 0.001	dbSNP: C: 99.822% (2250 / 2254); T: 0.178% (4 / 2254), EVS (AA) A=50/G=3886
rs2236178	5	c.707T>C	148	p.Met236Thr	T=0.34	dbSNP: C: 64.369% (3526.800 / 5479); N: 0.018% (1 / 5464); T: 35.612% (1951.200 / 5479), **EVS** (EA)

			heterozygous /153 homozygous			C=5702/T=2592 (AA) C=2660/T=1280
Not known	5	c.709C>A	heterozygous	p.Pro237Thr	A= 0.001	not known SNP
rs35460260	5	c.929A>T	heterozygous	p.Glu310Val	T= 0.001	dbSNP: A: 0.444% (10 / 2254); T: 99.556% (2244 / 2254) , **EVS** (AA) G=51/C=3695
rs151319642	5	c.1024C>G	heterozygous	p.Pro342Ala	G= 0.006	dbSNP: C: 99.770% (2173 / 2178); G: 0.230% (5 / 2178), **EVS** (AA) G=51/C=3695
Not known	6	c.1061A>G	heterozygous	p.E354G	G= 0.001	not known SNP
rs80251208	6	c.1331C>G	heterozygous	p.Ser444Cys	G= 0.001	No frequency in dbSNP or 1000Genome, found in Exome chip, Illumina, NHLBI-ESP , **EVS** (AA) G=26/C=3648
Not known	7	c.1383G>A	heterozygous	p.(=)	A= 0.001	not known SNP
rs36064635	8	c.1488C>T	heterozygous	p.(=)	T= 0.001	dbSNP: A: 0.444% (10 / 2254); G: 99.556% (2244 / 2254), **EVS** (EA) T=1/C=8209 (AA) T=83/C=3679
rs201638838	10	c.1745G>A	heterozygous	p.Arg582Lys	A= 0.003	No frequency in dbSNP or 1000Genome, found in Exome chip, NHLBI-ESP , **EVS** (AA) A=5/G=3805
rs34011504	11	c.1809C>T	heterozygous	p.(=)	T= 0.003	dbSNP: A: 0.975% (22 / 2256); G: 99.025% (2234 / 2256), **EVS** (EA) T=1/C=8305 (AA) T=104/C=3792
rs34269420	11	c.1870C>T	heterozygous	p.Pro624Ser	T= 0.006	dbSNP: A: 0.886% (20 / 2256); G: 99.114% (2236 / 2256), **EVS** (AA) T=106/C=3656
rs115272132	IVS12	c.1924+3G>A	heterozygous	…..	A= 0.004	dbSNP: A: 0.523% (12 / 2296); G: 99.477% (2284 / 2296), **EVS** (EA) A=1/G=8155 (AA) A=80/G=3544

Table S1: Benign *KIZ* variants identified in autosomal recessive RCD subjects: Ref.: NM_018474.4