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Supplemental Data

Homozygous Mutations in *PXDN* Cause Congenital Cataract, Corneal Opacity, and Developmental Glaucoma

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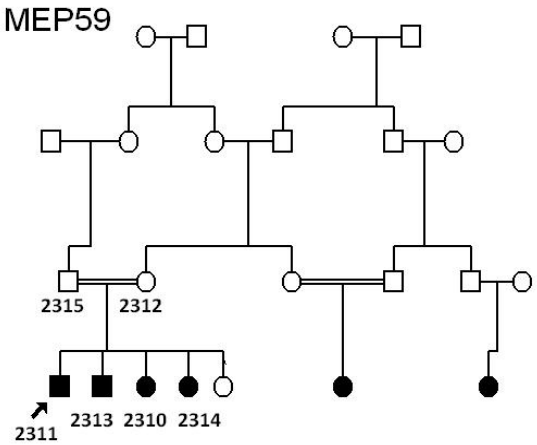

Marker	Physical distance (Mb)	MEP59						LOD SCORE
		2310	2311	2312	2313	2314	2315	
		Affected	Affected	Unaffected	Affected	Affected	Unaffected	
D2S2268	0.2	218 218	218 218	218 218	218 218	218 218	218 218	2.10
D2S323	2.1	189 189	189 189	189 189	189 189	189 189	187 189	2.31
D2S2245	2.8	172 172	172 172	168 172	172 172	172 172	172 172	1.74
D2S319	3.4	119 119	119 127	119 119	119 119	119 119	119 127	-2.84
D20S103	0.6	97 99	93 95	93 99	93 95	97 99	95 97	-10.73
D20S193	3.3	149 153	151 153	149 151	145 151	149 153	145 153	-10.73
D20S482	4.5	153 161	153 153	153 161	153 157	153 161	153 157	-9.56
D20S895	5.1	214 222	214 216	216 222	216 216	214 216	214 216	-8.73

Figure S1. Homozygosity Mapping in Family MEP59
Microsatellite genotyping data for chromosomes 2p and 20p are shown. The physical distance for each marker is based on the human February 2009 assembly (hg19) of the UCSC Genome Browser. Allele sizes are shown in base pairs. Note that the chromosome 2p region is homozygous in each of the affected members (highlighted in grey). A maximum multipoint LOD score of 2.31 was calculated at D2S323 using the SUPERLINK program with 0.001 mutant allele frequency and 100% penetrance under a recessive model.

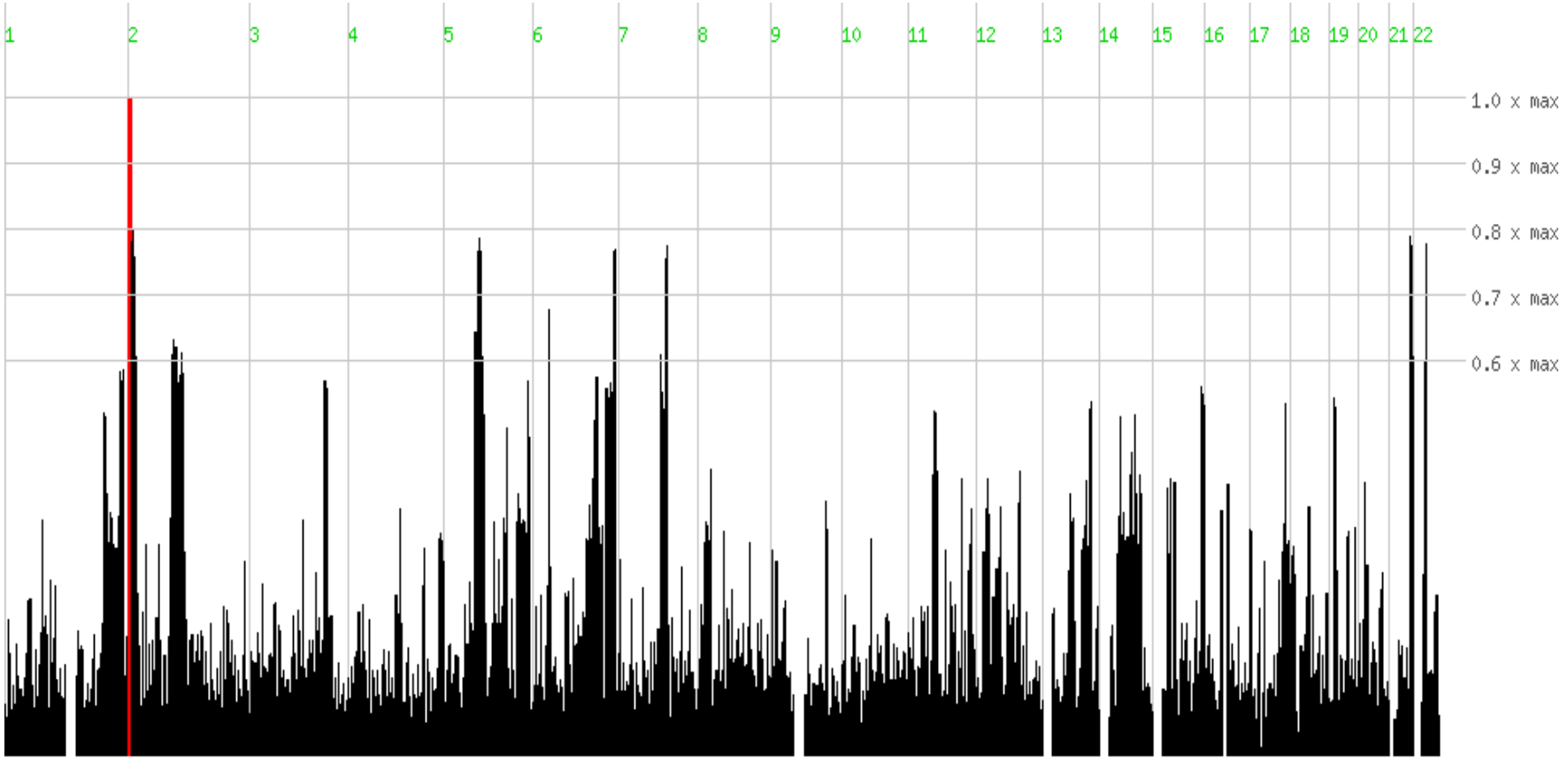

Figure S2. Homozygosity Mapping in Family CA2

Genomic DNA from each of the four affected individuals was genotyped on HumanHap610 Quad Arrays (Illumina, San Diego, CA, USA) at the Australian Genome Research Facility, Melbourne, Australia, according to standard protocols. Homozygosity Mapper output highlighted a single significant peak indicated in red at the p telomere of chromosome 2 that was common to the four affected individuals in this family.

Table S1. Summary of Output following Targeted Capture and Next Generation Sequencing for MEP60 (and the 7 Unrelated DNAs)

CHROM	POS	REF	MEP60	QUAL	DNA1	DNA2	DNA3	DNA4	DNA5	DNA6	DNA7	Gene	Function	cDNA	Protein
chr2	1652983	AG	A	1299.31	AG	AG	AG	AG	AG	AG	AG	PXDN	frameshift deletion	c.2568delC	p.P856fs
chr20	2413163	G	C	2232.61	G	G	G	G	G	G	G	TGM6	nonsynonymous SNV	c.1995G>C	p.R665S
chr20	1902349	A	A/G	260.23	A/G	A	A	A	A	A	A	SIRPA	nonsynonymous SNV	c.745A>G	p.T249A
chr20	3766941	T	T/G	268.7	T/G	T	T	T/G	T/G	T	T/G	CENPB	nonsynonymous SNV	c.190A>C	p.T64P
chr20	5454332	T	T/C	275.65	T	T	T/C	T/C	T/C	T/C	T/C	LOC643406	nonsynonymous SNV	c.44T>C	p.L15P
chr20	1552348	GGT	G	1831.27	GGT/G	GGT	GGT	GGT	GGT	GGT	GGT	SIRPB1	intronic	-	-
chr20	5548208	TAA	T	987.5	T	TAA/T	T	TAA	TAA	T	TAA/T	RP5-1022P6.2	intronic	-	-

The variants remaining after filtering against dbSNP131 are summarised. The chromosome (CHROM), position from the top of the chromosome in bp (POS), reference sequence at this position (REF), MEP60 sequence at this position (MEP60), quality of the sequence variant at this position (QUAL), the sequence at this position for the 7 unrelated DNAs (DNA1-7), Gene ID, function of the variant, cDNA and protein sequence nomenclature for the variation are depicted.

Table S2. Human *PXDN* Oligonucleotide Primers

Exon	Primer Sequence (F)	Primer Sequence (R)	Product Size (bp)
1	AGTCTCGAGCGCAGAATCAG	TCCCGGATCTCCACGAAG	557
2	AGCAAATGTGTAGCGATGTTTG	CCACTGATTGTGGATGATGAG	230
3	TGTCAATGTGACCTTTTCCAAG	ACAATAATTCATAAGCAGCACG	265
4	CTTTTCCGTCTGAAGTTTGACC	CATGCAAACAGCTAGCTCACTC	230
5	AATTTGGGCAAAAATCTGGA	GAATGGAAAGGCAAGGAACA	176
6	CCATTTCTTTTGTTTTCAGATTCA	AGAGAGAAAGAAGGCTTTGCAG	277
7	GCTCAGATTCCACCACGAGAC	CACTCACCACAGTTCAGCTCTT	334
8	TCTGTGGTTTCCTTTTGGAC	GTGTGGTTTGGGTGTCTGAG	289
9	ATGGGTGAGGTACGTCTTTGTG	ATTCTTATTTTGGGGTGCAAGG	323
10	AAAACCATGTCAAACCGGGA	GACAGGGCAGAAGATTCTATGG	436
11	GACTGTGGTCATTTTCTTGGTC	AGTGGAAATGTGTGCTTCCTG	267
12	TCTGTGGTCATGCTGTCAGTC	GTAACACTAATGTCAGCTGCGG	327
13	GGCAGGGGATTTTGTTCTTC	CCAGCCCGTCTACCTTGTATC	246
14	CCCCAACCCTAGGTCAGATAAC	CTGGGACCACACTAGGGACC	306
15	GTATGCCTTGTGGCTAATGATG	TTTAATCACTCCCACCTTCACC	275
16	AATAGCAATGATTATATTTTGGGG	AATGTGACTTAACTGAGGCGTG	322
17A	ATTGAGTGCGTCTTGCTTTTCT	CATCAGCATGTGGGTGAACT	386
17B	GTGTACGAGAATGGCTTCAACA	GGTACACGGAGTTCATGAGCAG	388
17C	GCTGCTCATGAACTCCGTGT	GCGTGTGCATGCTGGTCAGG	296
17D	CGAGAACGAGAGCCCCATCC	CGTCCAGCCGGTAAAGCAGT	371
17E	ATCAATGCTGGCATCTTCAAC	CCTAGGTACACGAGCCATCC	481
18	CACGTGCAGCTTTGTTAGGTAG	AGTCTCAACCAGGACAGCTCTT	269
19	TAGTGACCTGTGAGTCTCCTGC	TGACATCTGCAGGTCATTAAGC	354
20	TGGCAGGATGTGCTTAATTTC	CAGGATGCCGGTCCTACTGC	273
21	CGTGACCTGCCAAGGTTATATT	ACAGTTGCCTGGGAATAAAATG	285
22	CTTTTAATACAAGAGCCAAGCTG	ACATGGGACTCTCGGACTTG	244
23	AGCTGACAGGCTTTGCTCAC	AGTTCTGGGTGTTTCCTGGTC	291

Primer pairs used for the amplification of the coding region and splice site junctions of human *PXDN*.