

Supplemental Data

A Homozygous Mutation in the Tight-Junction Protein

JAM3 Causes Hemorrhagic Destruction of the Brain,

Subependymal Calcification, and Congenital Cataracts

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Table S1. Blocks of homozygosity over 1.0cM shared by the affected individuals tested

Block#	Chromosome	#Markers in the block	Block size (cM)	Block size (Mb)	Proximal boundary	Distal boundary
1	11	1039	8.5	3.2	rs1627835	rs1893054
2	11	72	1.3	0.5	rs2241574	rs4937504
3	1	2	1.2	0.8	rs4845917	rs2933080
4	14	2	1.1	0.7	rs11628672	rs8015048
5	10	4	1.0	1.6	rs3802542	rs747421
6	14	38	1.0	0.3	rs1677999	rs11160674

Table S2. Summary of candidate gene sequencing

Gene symbol	MIM no.	Sequencing results
<i>OPCML</i>	600632	No mutation found
<i>SPATA19</i>	609805	No mutation found
<i>IGSF9B</i>	Not available	No mutation found
<i>JAM3</i>	606871	Splice site mutation found
<i>NCAPD3</i>	609276	No mutation found
<i>VPS26B</i>	610027	No mutation found
<i>THYN1</i>	Not available	No mutation found
<i>ACAD8</i>	604773	No mutation found
<i>GLB1L3</i>	Not available	No mutation found

Table S3. Primers used for amplification and sequencing of *JAM3* coding sequence

Amplicon name	Forward primer (5'>3')	Reverse primer (5'>3')	Length of PCR product (bp)
<i>JAM3</i> Exon 1	TTCCTTCTCCGACCTGTTGT	GGAGCCAGGACCGCTATC	497
<i>JAM3</i> Exon 2	CTCTTTCCCTTGGCTCCTTC	GAGTTGGCTGCAAGACATCA	346
<i>JAM3</i> Exon 3	CTTGGAGTGTGGAGTTGGCT	ACTGCACTTGTTGATGCTGG	477
<i>JAM3</i> Exon 4	CCAGAAACCACCCTCTTCAA	TCAGGCAGTCTCAGCTCTCA	373
<i>JAM3</i> Exon 5	TGGAGCTGATTTCTTCTGGG	GGAAGAAGGGTCATTTTTCTG	443
<i>JAM3</i> Exon 6	GGAGCTGTCAGGCAGGTATC	TTTTCTCAGCCTTCCCTTCA	441
<i>JAM3</i> Exon 7	TCTCTTCTAGCTGGGGTAGGC	AAGGGGGAATGTTGTTTTCC	473
<i>JAM3</i> Exon 8	GTA CTGGCCCTGATCACGTT	GAAATCTTCCCAGGAGACCC	315
<i>JAM3</i> Exon 9	GCCCATGTTAGACTTACTCCG	GACCAGTGGTGAACATGCTG	481

Table S4. Primers used for amplification and sequencing of *JAM3* cDNA to determine splicing of intron 5

Forward primer (5'>3')	Reverse primer (5'>3')	Length of PCR product (bp)
TTATCGCTGTGAGGTCGTTG	CGCCAATGTT CAGGTCATAG	395

The forward primer is located within exon 4, and the reverse primer spans across exon 6 and exon 7.