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Supplemental Data

Mutations in *LAMA1* Cause Cerebellar Dysplasia and Cysts with and without Retinal Dystrophy

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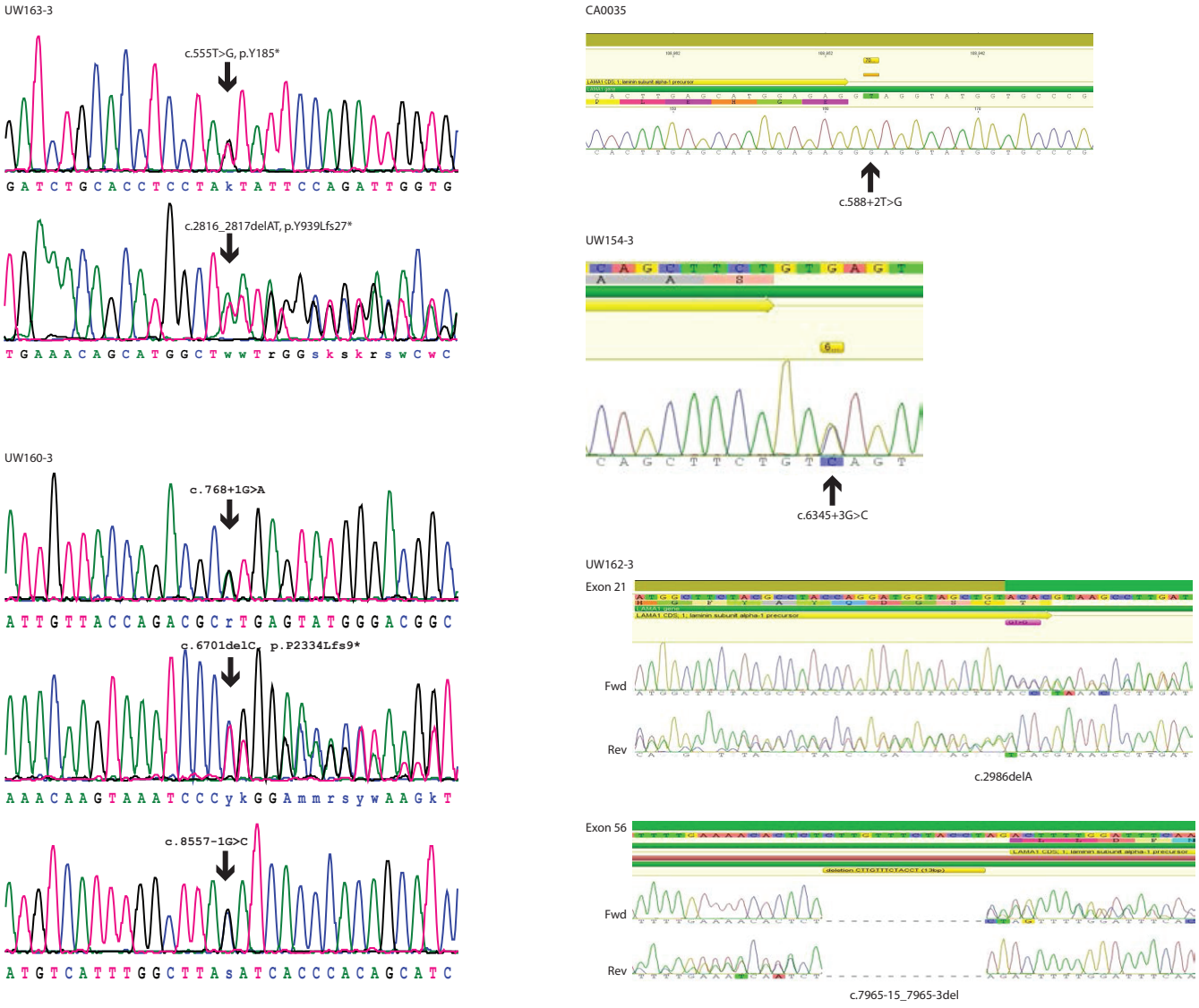

Figure S1. Sanger validations. Chromatograms showing Sanger sequencing confirmation for variants identified through exome sequencing in probands.

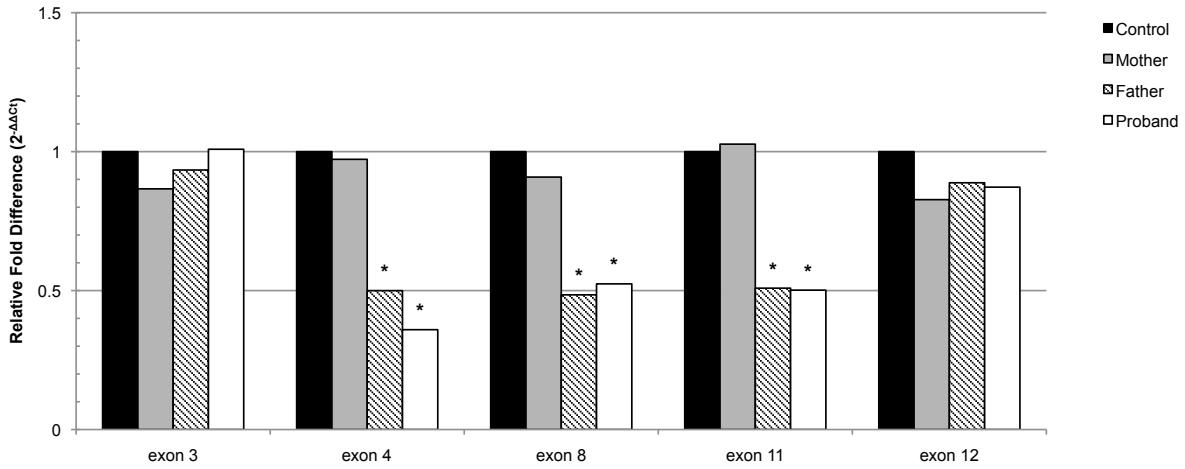

Figure S2. qPCR confirmation of paternally inherited *LAMA1* exon 4-11 deletion in UW154-3. Relative comparison of exon copy number within and flanking the deleted region. Fold-differences were calculated using the comparative-Ct method relative to a pooled sample of 1,000 unaffected males. Asterisk denotes copy number loss (0.5 fold differences relative to control).

Table S1. QC metrics and coverage summary of exome data in affected individuals with *LAMA1* mutations

	CA0035	UW154-3	UW162-3	UW160-3	UW163-3
Total reads	127 385 980	113 569 944	118 968 257	93 983 564	122 559 090
Total aligned reads	122 364 705	109 355 622	116 472 257	93 839 687	122 339 041
Mean read length	96.72	94.5	94.661	49.1	49.45
Mean coverage	130.96X	111.38X	126.83X	51.58X	66.1X
Coverage in CCDS region					
5X	97.60%	97.70%	97.70%	95.00%	95.30%
10X	96.90%	97.00%	97.10%	91.40%	92%
20X	95.10%	95.10%	95.30%	82.00%	83.90%
50X	84.80%	82%	84.30%	46.30%	56.40%

Table S2. Variant filtering summary of exome data in affected individuals with *LAMA1* mutations

	CA0035	UW154-3	UW162-3	UW160-3	UW160-3
Total variants	220 838	201 040	216 288	121 246	151 142
Non-synonymous/splicing/coding indel variants	13 401	11 991	11 758	11 218	12 908
After excluding variants reported in 1000 genomes and EVS datasets (frequency >0.5%)	615	504	427	346	1 285
After excluding variants present in >5 in-house exomes	395	279	225	306	1 132
Homozygous variants in CA0035	19	---	---	---	
Genes shared by all affected	TTN, LAMA1				