

## Supplement

**Table S1)** List of primer sequences used for qPCR

Gene	Forward (5'-3')	Reverse (5'-3')
<i>ATP9A</i>	ACCATGCAGGCTGTCTTTTC	TCAGGATACAGCATGGCAAC
<i>ARPC3</i>	GCCCCCAGAGAGACAAAAG	TTGGAATTGCACTTTTGCAG
<i>SNX3</i>	CAGGGTCAAGACAAATCTTCC	GGATGACCAGCGACCTTG
<i>GAPDH</i>	TGCACCACCAACTGCTTAGC	GGCATGGACTGTGGTCATGAG

**Table S2)** List of primer sequences used for cDNA sequencing of *ATP9A*

Forward (5'-3')	Reverse (5'-3')
TTCCTGGGGTGCTGTTCAA	AGTACCTGATGCGACCACAG

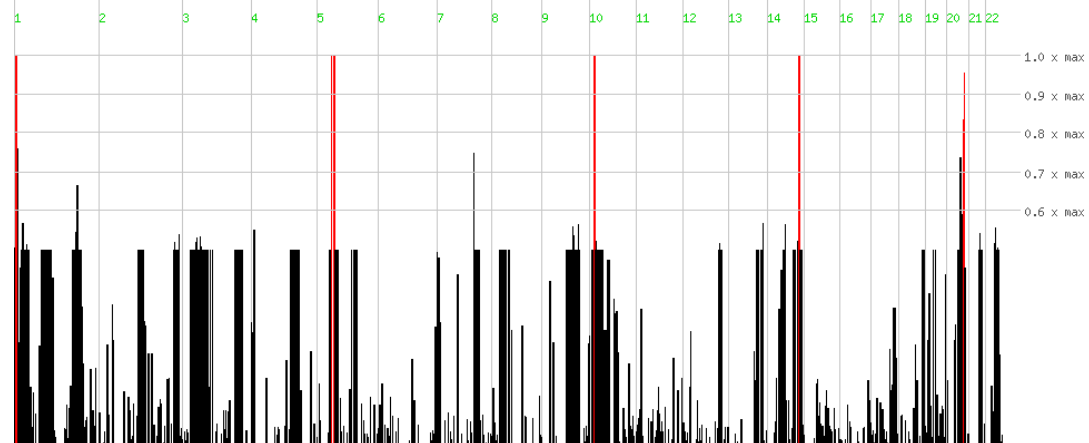
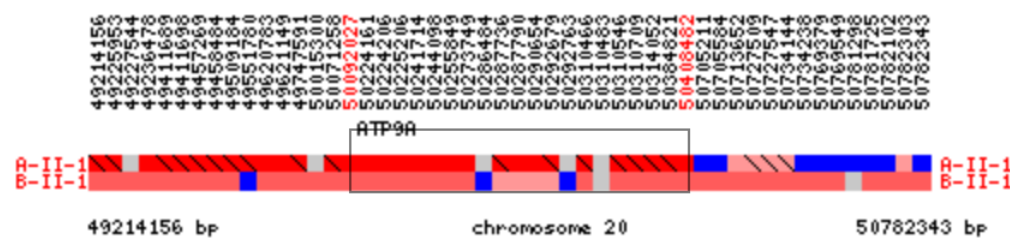
**Table S3)** Splice Prediction scores indicating loss of authentic donor site

Splice Prediction score	Score
SpliceAI (Delta score for Donor Loss)	0.99
MaxEntScan	8.56
ada score	0.99
dbscSNV rf score	0.91

**Figure S1)**

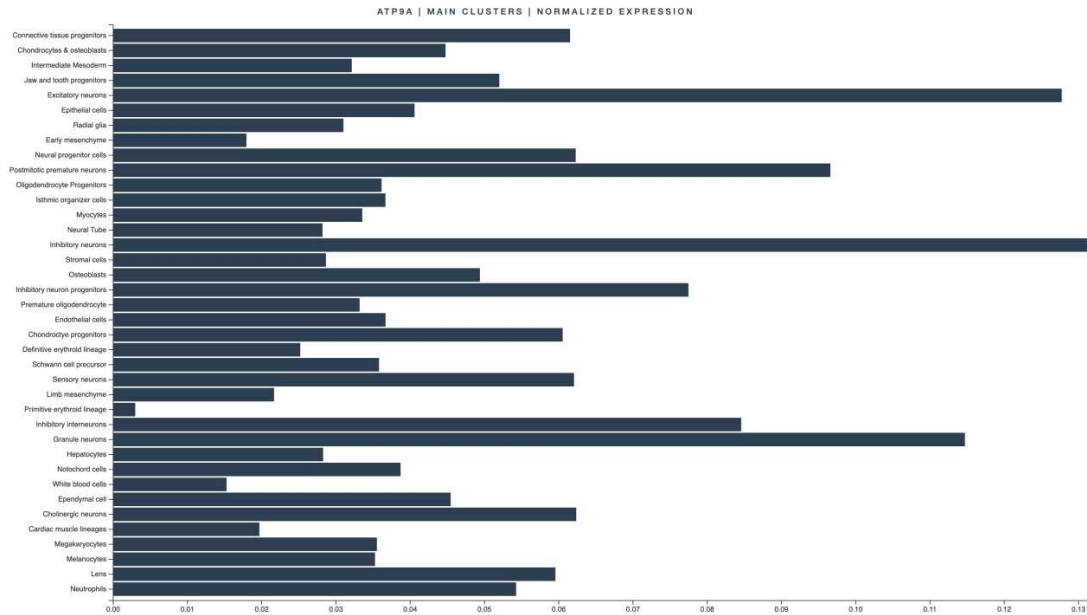
These screenshots from AutozygosityMapper (<https://www.genecascade.org/AutozygosityMapper/>) show the genome-wide sharing of homozygous segments (S1a) and the underlying genotypes (S1b) around the *ATP9A* locus in the two affected individuals for which WES data were available (red: homozygous genotypes; blue: heterozygous genotypes; grey: low coverage). It should be noted that the two patients do not have the same haplotype, as shown by diagonal bars indicating different alleles.

## S1a) genome wide homozygosity

S1b) genotypes at the *ATP9A* locus

**Figure S2)**

Screenshot from MOCA <https://oncoscape.v3.sttrcancer.org/atlas.gs.washington.edu.mouse.rna/genes> showing strong *Atp9a* expression in different neuronal cells measured via sci-RNA-seq3



**Figure S3)**

Screenshot from <https://gtexportal.org/home/gene/ATP9A> showing total *ATP9A* expression

