

Supplemental Data

Mutation of the Variant Alpha Tubulin TUBA8

Results in Polymicrogyria with Optic Nerve Hypoplasia

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Neuroimaging, individual VII:1 Family 1

Figure S1. T1 sagittal section in the midline. The corpus callosum is thinned and deficient posteriorly (white arrow). The cingulate gyrus is poorly formed and the cerebral gyri are disorganised. The brainstem is abnormal with loss of demarcation of the pontomedullary junction (yellow arrow).



Figure S2. T2 axial image above the level of the ventricles. There is very extensive polymicrogyria extending bilaterally throughout the brain, including in the parafalcine regions (arrowed).

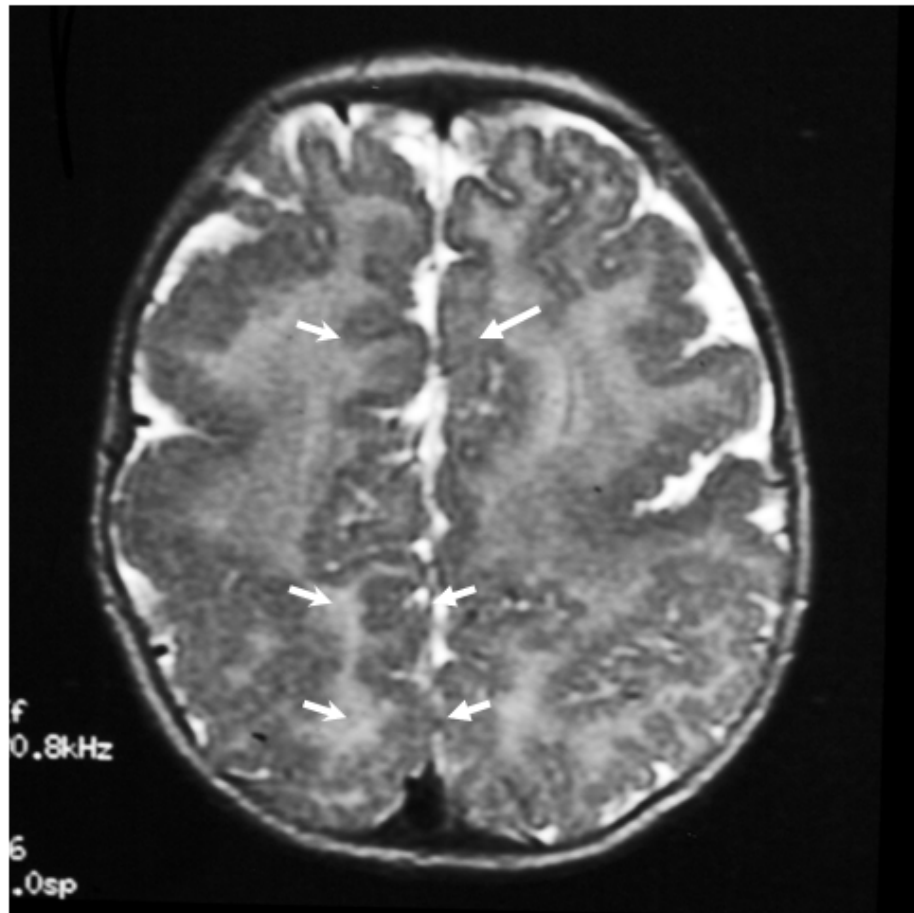
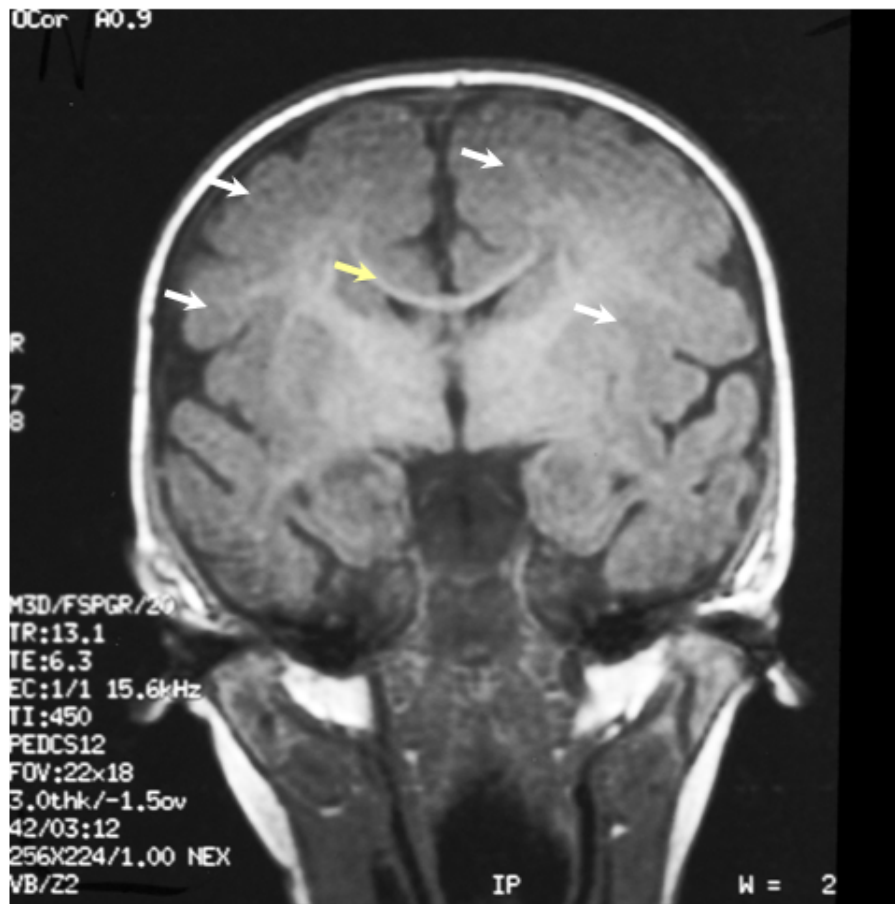


Figure S3. Coronal T1 image at the level of the third ventricle. The thin corpus callosum is apparent (yellow arrow). There is thickening of the cortical grey matter (white arrows).



Neuroimaging, individual VII:5 Family 1 (images are affected by movement artefact)

Figure S4. T1 sagittal section through the midline. The corpus callosum is largely absent and the cingulate gyrus is deficient (yellow arrow). The cerebral gyral pattern is abnormal. There is an abnormality of the brainstem with loss of demarcation of the pontomedullary junction (white arrow). This results in a thickened medulla in the antero-posterior dimension.



Figure S5. T2 axial section above the level of the ventricles. The cortex is thickened and the gyral pattern is abnormal. There is extensive bilateral polymicrogyria extending both anteriorly and posteriorly (arrows).

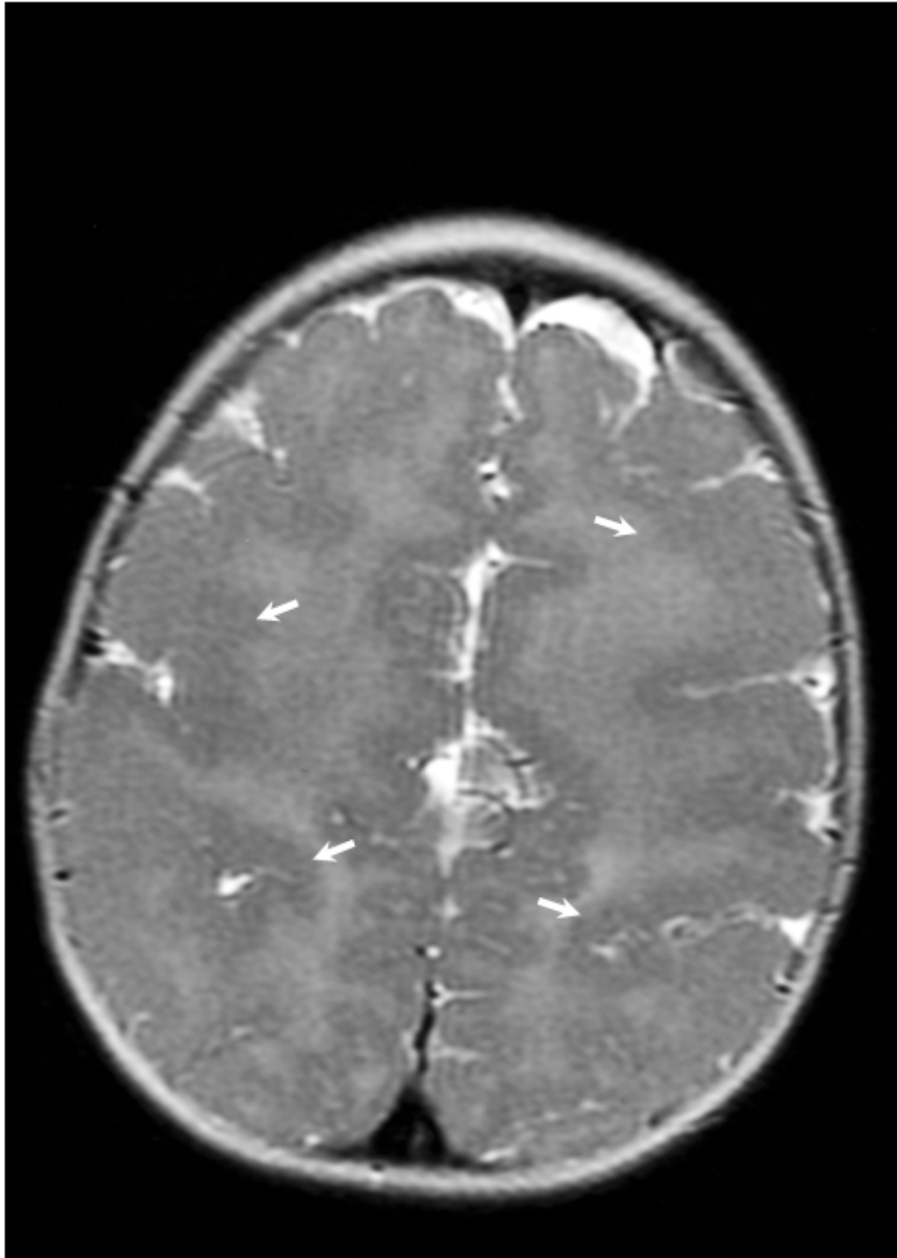
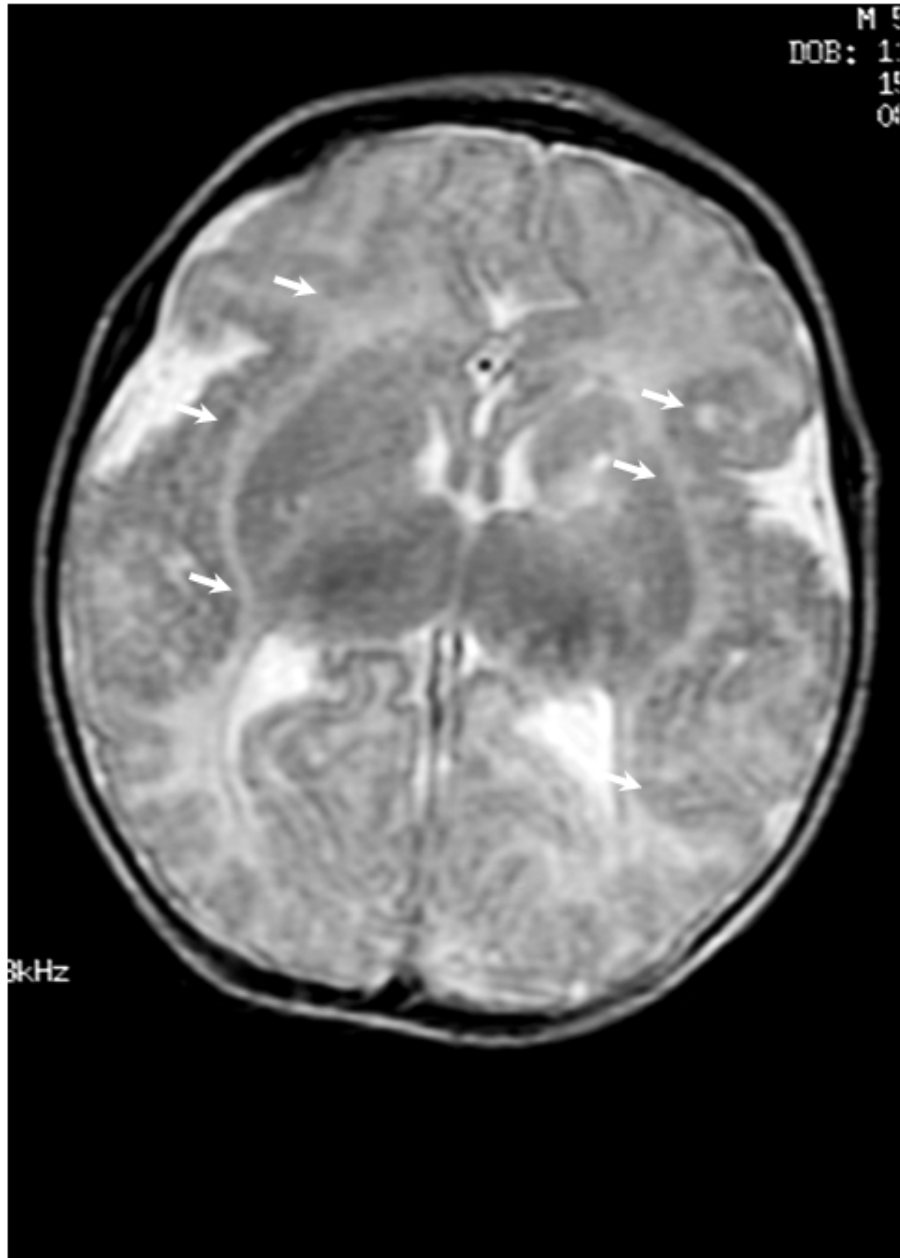


Figure S6. T2 axial section at the level of the basal ganglia, showing the absence of the corpus callosum. There is very extensive polymicrogyria bilaterally extending throughout the brain from anterior to posterior (arrows)



Accession Numbers

Tuba8 Genomic reference sequence: NC_000022; mRNA reference sequence: NM_018943.

Table S1. PCR Primers Used for Sequencing and RT-PCR

Primers used for sequencing of <i>TUBA8</i>		
Exon 1F	5' - GGTCACGAAGGCTGCTCTC - 3'	
Exon 1R	5' - GACTCCTCGACCCTGAACTG - 3'	
Exon 2F	5' - GCTATAGAGCTGGGGCACCT - 3'	
Exon 2R	5' - GTTGCACCATCCCCACCT - 3'	
Exon 3F	5' - GAGGTGGTCTGGCTTCAAAC - 3'	
Exon 3R	5' - TGATGCCAACACAGAGGAAG - 3'	
Exon 4Fi	5' - GCTGTTTTGATTTTCATCCACAA - 3'	
Exon 4Ri	5' - GCTTTCTCGGCAGAGATGAT - 3'	
Exon 4Fii	5' - ACCGCCTCATCAGTCAGATT - 3'	
Exon 4Rii	5' - CAGCACTTGTGTCTCAACCAA - 3'	
Exon 5F	5' - ATCCCATAGCCCCACTCC - 3'	
Exon 5R	5' - GTCCACTTAGCCCAGTCGTG - 3'	
Primers used for analysis of LCL <i>TUBA8</i> transcripts by RT-PCR		
Forward	5' - CGGCTGTATCTGGAGCAGTC - 3'	
Exon 2 Reverse	5' - GGCTCCAGATCTATCATGACG - 3'	
Exon 3 Reverse	5' - CTTCCGTATGCGGTCCAGCA - 3'	
Exon 4 Reverse	5' - TAGGTAGGGCGCTCAATGTC - 3'	
Exon 5 Reverse	5' - CAAACGAATCAGTCCCCACT - 3'	