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Supplemental Data

Biallelic Mutations in *TMTC3,* Encoding a Transmembrane and TPR-Containing Protein, Lead to Cobblestone Lissencephaly

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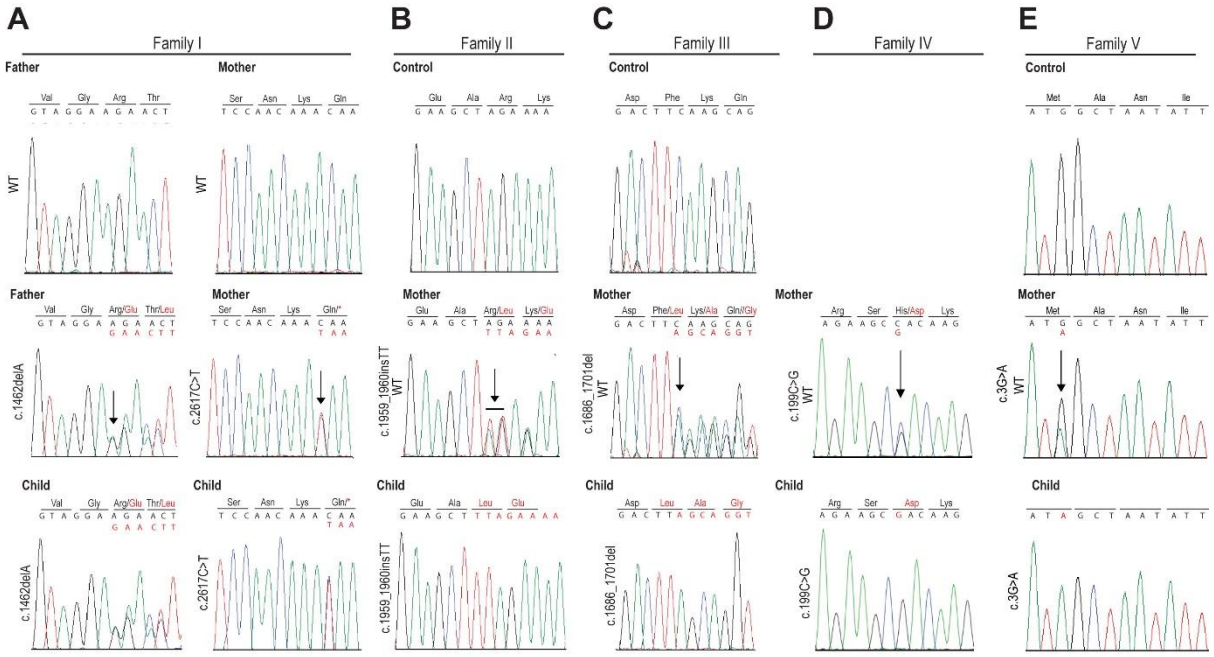
Figure S1. *TMTC3* variants in families with Cobblestone lissencephaly.



(A) Chromatrograms of exons 11 and 14 showing heterozygous 1bp A deletion in exon 11 from the father and the C to T substitution resulting in a premature stop codon mutation from the mother in family 1. (B) Chromatograms of exon 14 showing a TT insertion in family II. (C) Chromatograms of exon 12 showing a 30 bp deletion in family III. (D) Chromatograms of exon 3 showing a C to G substitution observed in family IV. (E) Chromatograms of exon 2 showing a G to A substitution observed in family V.

Table S1. Summary of the pathogenic *TMTC3* (*NM_181783.3*) variants detected in this study.

Family	Individual	Zygosity	Genomic position ch12	Exon affected	Nucleotide variant	Amino acid alteration	Type of mutation
I	1	Heterozygous	88582647	11	c.1462delA	p.Arg488Glufs*6	Frameshift
		Heterozygous	88589298	14	c.2617C>T	p.Gln873*	Stop
II	2	Homozygous	88588639	14	c.1959_1960insTT	p.Arg654Leufs*6	Frameshift
II	3	Homozygous	88588639	14	c.1959_1960insTT	p.Arg654Leufs*6	Frameshift
III	4	Homozygous	g.88584379_88584394del	12	c.1686_1701del	p.Phe562Leufs*8	Frameshift
IV	5	Homozygous	88547077	3	c.199C>G	p.His67Asp	Missense
IV	6	Homozygous	88547077	3	c.199C>G	p.His67Asp	Missense
V	7	Homozygous	88542095	2	c.3G>A	p.Met1?	Initiation defect
V	8	Homozygous	88542095	2	c.3G>A	p.Met1?	Initiation defect
VI	9	Heterozygous	88566474	8	c.1151G>A	p.Gly384Glu	Missense
		Heterozygous	88589194	14	c.2521dupA	p.Ile841Asnfs*4	Frameshift