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Supplemental Data

Mutations in *DPAGT1* Cause a Limb-Girdle Congenital Myasthenic Syndrome with Tubular Aggregates

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Ethical approval for studies on congenital myasthenic syndromes was obtained from OXREC B: 04.OXB.017 and Oxfordshire REC C 09/H0606/74. The approvals include research and screening of DNA provided that this research and screening directly relates to congenital myasthenic syndromes.

Table S1. Analysis of Whole-Exome Sequencing Data

	Number of Variants			
	Case 1	Case 2	Shared between Two Cases	Shared between Two Cases, but Absent from Other In-House Exomes
All variants after filtering using dbSNP132 database	1,574	1,287		
Nonsynonymous changes, splicing mutations, changes in 3'UTRs and 5'UTRs	377	300		
Genes with two or more compound heterozygous mutations or with homozygous mutations	98	69	34	1 - *DPAGT1*

Table S2. Predicted Pathogenicity of *DPAGT1* Mutations

	DNA Mutation	Protein Alteration	Segregation in Family Members	PolyPhen2 Prediction	PolyPhen2 Score
Case 1	c.324G>C	p.Met108Ile	Individual trans-heterozygous based on exome sequencing.	probably damaging	0.957
	c.349G>A	p.Val117Ile	Individual trans-heterozygous based on exome sequencing.	possibly damaging	0.802
Case 2	c.349G>A	p.Val117Ile	One heterozygous unaffected sibling.	possibly damaging	0.802
	c.699dup	p.Thr234Hisfs*116	Two heterozygous unaffected siblings.	frameshift insertion	
Case 3	c.358C>A	p.Leu120Met	Heterozygous unaffected parent, one compound heterozygous affected sibling (Case 5).	probably damaging	1
	c.791T>G	p.Val264Gly	Heterozygous unaffected parent, one heterozygous unaffected sibling, one compound heterozygous affected sibling (Case 5).	probably damaging	1
Case 4	c.478G>A	p.Gly160Ser	N/A	probably damaging	1
	c.574G>A	p.Gly192Ser	N/A	probably damaging	1
Case 5	c.358C>A	p.Leu120Met	Heterozygous unaffected parent, one compound heterozygous affected sibling (Case 3).	probably damaging	1
	c.791T>G	p.Val264Gly	Heterozygous unaffected parent, one heterozygous unaffected sibling, one compound heterozygous affected sibling (Case 3).	probably damaging	1