

Table S1: CMS subtype-specific serum creatine kinase levels within the Oxford CMS Cohort

CMS subtype	CMS gene	n	CK range (IU/L) <200 = normal	Mean \pm SD
<i>Glycosylation</i>				
ALG-2/14	<i>ALG-2, ALG14</i>	6	38-269	145.0 \pm 99.4
DPAGT1	<i>DPAGT1</i>	4	69-146	106.2 \pm 33.7
GFPT1	<i>GFPT1</i>	4	95-1731	596.7 \pm 769.0
GMPPB	<i>GMPPB</i>	6	418-4807	2149.1 \pm 1452.9
<i>Presynaptic</i>				
Choline acetyltransferase	<i>CHAT</i>	1	49	-
<i>Synaptic</i>				
Agrin	<i>AGRN</i>	3	normal *	-
Endplate AChE deficiency	<i>COLQ</i>	12	14-64	43.0 \pm 21.2
<i>Postsynaptic</i>				
DOK7	<i>DOK7</i>	22	19-672	148.3 \pm 152.9
Fast channel syndrome	<i>CHRNA, CHRNB, CHRND, CHRNE</i>	3	119, normal *	-
MuSK	<i>MUSK</i>	1	54	-
Primary AChR-deficiency	<i>CHRNA, CHRNB, CHRND, CHRNE</i>	41	6-258	109.3 \pm 60.7
Rapsyn	<i>RAPSN</i>	18	18-295	93.7 \pm 65.0
Slow channel syndrome	<i>CHRNA, CHRNB, CHRND, CHRNE</i>	12	51-376	154.4 \pm 126.7

* Specific CK values were not available, but contemplated as normal in the patients' notes.