UKGTN Testing Criteria

Test name: **Myasthenia, Limb-Girdle, With Tubular Aggregates Type 2 And Congenital Disorder Of Glycosylation, Type Ij**	
Approved name and symbol of disorder/condition(s): Myasthenic Syndrome, Congenital, with Tubular Aggregates 2; CMSTA2 Congenital Disorder of Glycosylation, Type Ij; CDG1J. **Approved name and symbol of gene(s):** dolichyl-phosphate (UDP-N-acetylglucosamine) N-acetylglucosaminephosphotransferase 1 (GlcNAc-1-P transferase); DPAGT1	**OMIM number(s):** 614750 608093 **OMIM number(s):** 191350

Patient name:	**Date of birth:**
Patient postcode:	**NHS number:**
Name of referrer:	
Title/Position:	**Lab ID:**

Referrals will only be accepted from one of the following:	
Referrer	**Tick if this refers to you.**
Consultant clinical geneticist	
Consultant neurologist	
Consultant paediatric neurologist	

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:	
Criteria	**Tick if this patient meets criteria**
Congenital Myasthenic Syndrome with a limb-girdle pattern of weakness **AND** at least one of the following: • anti-cholinesterase responsive • tubular aggregates on muscle biopsy	
OR at risk family members where familial mutation is known	

Additional Information:
If the sample does not fulfil the clinical criteria or you are not one of the specified types of referrer and you still feel that testing should be performed please contact the laboratory to discuss testing of the sample.