UKGTN Testing Criteria

Test name: **Rhabdomyolysis and Metabolic Myopathies 30 Gene Panel**	
Approved name and symbol of disorder/condition(s): See website listing	**OMIM number(s):**
Approved name and symbol of gene(s): See website listing	**OMIM number(s):**

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 Paediatrician	
Consultant Neurologist	
Consultant in Metabolic Medicine	
Consultant Clinical Geneticist	

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:	
Criteria	**Tick if this patient meets criteria**
Myalgia, muscle weakness, cramps, myoglobinuria, triggered by exercise/heat/fasting/infection **AND**	
Suggestive blood/urine tests: acylcarnitines, lactate, amino acids, organic acids, Creatine Kinase (CK) **AND**	
Mitochondrial myopathy/dystrophinopathy unlikely/excluded **AND**	
Common McArdle disease (*PYGM*)/CPTII deficiency (*CPT2*) mutations excluded, as appropriate	

Additional Information:
See testing pathway over the page.

For panel tests: At risk family members where familial mutation is known do not require a full panel test but should be offered analysis of the known mutation

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.

Approval Date: Sept 2015