UKGTN Testing Criteria

Test name: Disorders associated with Hyperammonaemia & Fatty Acid Oxidation and Disorders of Ketogenesis or Ketolysis 38 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 Clinical Geneticist	
Consultant Neurologist (Adult or Paediatric)	
Consultant Metabolic Specialist (Adult or Paediatric)	

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:	
Criteria	**Tick if this patient meets criteria**
Abnormal clinical features including encephalopathy, severe vomiting or loss of consciousness and one of the following:	
• Plasma ammonia >150µmol/L **OR**	
• Biochemical testing results indicative of fatty acid oxidation **OR**	
• Hypoketotic hypoglycaemia or severe ketoacidosis	

Additional Information:
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.