UKGTN Testing Criteria

Test name: **Hyperammonaemia/Urea Cycle Disorders 14 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:**

Referrer	**Tick if this refers to you.**
Consultant in Metabolic Disease (Paediatric/Adult)	
Consultant Paediatrician	
Consultant Neonatologist	
Consultant Hepatologist	
Consultant Neurologist	
Consultant Clinical Geneticist	

Criteria	**Tick if this patient meets criteria**
Hyperammonaemia (plasma ammonia >150µmol/L) **AND** at least **TWO** of the following: -	
1) Encephalopathy/cerebral edema	
2) Lethargy, loss of appetite, vomiting, behavioural abnormalfities, learning difficulties	
3) Plasma amino acids (esp citrulline, arginine)/urine orotic acid results suggestive of particular urea cycle defect: - Low citrulline, low arginine, low/normal urinary orotic acid - Low citrulline, low arginine, raised urinary orotic acid - Raised citrulline - Raised arginine	

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.