UKGTN Testing Criteria

Test name: **Carnitine Deficiency, Systemic Primary**	
Approved name and symbol of disorder/condition(s): Carnitine Deficiency, Systemic Primary; CDSP	**OMIM number(s):** **#212140**
Approved name and symbol of gene(s): solute carrier family 22 (organic cation transporter) member 5; SLC22A5	**OMIM number(s):** ***603377**

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 in Inherited Metabolic Disease	
Consultant Clinical Geneticist	

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:	
Criteria	**Tick if this patient meets criteria**
Patient with low levels of free carnitine, suspicion of having SCPD	
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.