

## UKGTN Testing criteria

**Name of Disease(s):** CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1C; CMT1C (601098)

**Name of gene(s):** Lipopolysaccharide-induced TNF factor; LITAF (603795)  
(also known as SIMPLE)

**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 UKGTN Gene Dossier:**

Criteria	Tick if this patient meets criteria*
1. CMT1 phenotype with median nerve MCV under or equal to 38m/sec; sensory impairment <b>AND</b>	
2. Negative molecular diagnosis of CMT1A, CMT1B, CMTX1, ie absence of mutation in <i>PMP22</i> , <i>GJB1</i> , <i>MPZ</i> , <i>EGR2</i> , <i>NEFL</i> , especially when patients present median nerve MCVs between 16 and 33 m/s <b>AND</b>	
3. Isolated case or pedigree suggestive for autosomal dominant inheritance	
Further details:	

\* If the patient 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.

Please return completed form to:

**Bristol Genetics Laboratory, Pathology Sciences,  
Southmead Hospital, Bristol BS10 5NB**