UKGTN Testing Criteria

Approved name and symbol of disease/condition(s):	**OMIM number(s):**
CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1D; CMT1D	607678
Approved name and symbol of gene(s):	**OMIM number(s):**
EARLY GROWTH RESPONSE 2; EGR2	129010

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.**
Clinical Geneticists	
Consultant Neurologists	
Consultant Paediatric Neurologists	

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:	
Criteria	**Tick if this patient meets criteria**
CMT1 or DSS (HMSNIII) presentation: • Severe early childhood onset degenerative distal demyelinating motor and sensory peripheral neuropathy. • CMT1 (nerve conduction velocity < 38 m/sec) ; or diagnosed DSS (with much slower NCV, and peripheral nerve hypertrophy /onion bulbs) **OR** diagnosed congenital hypomyelinating neuropathy (with hypotonia, delayed motor milestones, and absent myelination on biopsy)	
AND Isolated case or pedigree suggestive of autosomal dominant or autosomal recessive inheritance	
AND Exclusion of common forms of CMT1, CHN or DSD i.e. GJB1/MPZ/PMP22 negative	
At risk family members where familial mutation is known	

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.