

UKGTN Testing Criteria

Name of disease: Charcot Marie Tooth disease type 1; demyelinating
Name of gene: Early Growth Response 2 (EGR2)

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 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 CHN 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	

* 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**