AWMGS
All Wales Medical Genomics Service

Charcot-Marie-Tooth Disease Type 1A (CMT1A, Hereditary Motor and Sensory Neuropathy Type 1A, HMSN1A) – OMIM 118220

Background

CMT is a progressive condition that affects peripheral motor and sensory nerves – those in your legs and arms. The condition affects all ethnic groups equally across the world, and there are thought to be approximately 23,000 people with the condition in the UK. Symptoms vary greatly between each individual, even within the same family, but can include motor issues, leading to muscle weakness and wasting, and some sensory loss in the fingers, toes, legs or arms. Long term pain and chronic tiredness are the most common symptoms experienced by patients. CMT Type 1A is caused by a mutation (a duplication) in the gene *PMP22* (OMIM 601097) and follows an autosomal dominant inheritance pattern so a child has a 50% chance of inheriting the disease from an affected parent. Although this condition causes gradual deterioration of the nerves over time, it is not life threatening. Other genes and mutational mechanisms are responsible for the other types of CMT. Another type of neuropathy, Hereditary Neuropathy with Liability to Pressure Palsies (HNPP) is caused by a (deletion) mutation in the *PMP22* gene.

Recommended Clinical Referral Criteria

- Diagnostic cases and at-risk family members
- Progressive distal muscle weakness and atrophy, sensory loss and slow nerve conduction velocity
- Age of onset: 5 to 25 years

Molecular Analysis

Mutation screen: MLPA dosage analysis of *PMP22* gene to detect duplications
Family follow-up: Testing for known familial mutations in the *PMP22* gene

Test (Price available on request)	TAT
Diagnostic dosage analysis (*PMP22)*	42cd
Predictive analysis (*PMP22*)	28cd

Contact Details
All Wales Genomics Laboratory,
Institute of Medical Genetics,
University Hospital of Wales,
Heath Park,
Cardiff CF14 4XW
Tel: 029 2184 4023
Fax: 029 2184 4043
lab.genetics.CAV@wales.nhs.uk
medicalgenomicswales.co.uk
Accredited to ISO 15189:2012
(8988)

Sample Requirements
Blood – 5ml in EDTA (1ml neonates/infants);
Please contact lab prior to sending a prenatal sample.
Please label samples with three identifiers and date of collection
All samples must be accompanied by request form
Consent for testing & DNA storage is assumed when request for test received

Links

Orphanet
http://www.orpha.net/
OMIM
http://www.omim.org/
Genetic Test Registry
http://www.ncbi.nlm.nih.gov/gtr/

Support
http://www.cmt.org.uk/