East Midlands Regional Molecular Genetics Laboratory
City Hospital campus
Hucknall Road
Nottingham
NG5 1PB

Tel: 0115 9691169 ext. 55207
nuhnt.moleculargenetics@nhs.net
www.nuh.nhs.uk

Service information: Charcot Marie Tooth disease Type IA (CMT1A)

Gene/Locus: *PMP22* (17p11.2) **OMIM:** #118220

Alternative Name: Hereditary Motor and Sensory Neuropathy Type 1A (HMSN1A)
Referrals: Clinical Geneticists, Neurologists, Other Relevant Specialities
Testing: Diagnostic testing offered in clinically affected patients, presymptomatic testing in patients at risk of developing CMT1A (presymptomatic referrals are accepted from Clinical Genetics only)
Target Reporting Times:

- Diagnostic 28 calendar days
- Presymptomatic 14 calendar days From Clinical Genetics only

Please contact the laboratory if urgent testing is required

Test Details:

- **Copy number analysis of the *PMP22* region**, using multiplex ligation-dependent probe amplification (MLPA) (P033 Kit MRC Holland) to detect the common 1.5 Mb duplication of *PMP22*. This test will also detect the 1.5 Mb *PMP22* deletion associated with Hereditary Neuropathy with liability to Pressure Palsies (HNPP)

Service Details:

Approximately 70% of CMT type I cases have a duplication of the *PMP22* gene. Point mutations in other genes are also associated with CMT type I. *GJB1* (Connexin-32) testing is available in the East Midlands Regional Molecular Genetics Laboratory on request. DNA can also be sent to another centre to screen for mutations in a panel of genes associated with inherited peripheral neuropathies, but the cost of this testing is the responsibility of the referring clinician.

Sample Requirements:

- **EDTA** blood sample (1-4 ml), labelled with patient's **full name**, **date of birth** and **NHS number** or genomic DNA (50µl at ≥100ng/µl). Please send two separate blood/DNA samples for presymptomatic tests.
- Samples should be accompanied by a **fully** completed referral card which should include the patient's full name, date of birth and NHS number.
- Please also include details of the test, relevant clinical details and full details of the referring clinician and centre

Consent:

Please note that in submitting a sample, it is the responsibility of the clinician to ensure that consent has been taken i) for testing, ii) for storage, and iii) for the use of this sample and the information generated to be shared with the patient's relatives and their health professionals. Following testing, a sample of the patient's DNA may also be used anonymously to validate new tests and for internal quality control purposes.

Date modified – Feb 2016. If printed, this document is only valid on the day of printing