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Service information: Hereditary Neuropathy with Liability to Pressure Palsies (HNPP/HLPP)

Gene/Locus: *PMP22* (17p11.2) **OMIM:** #162500

Referrals: Clinical Geneticists, Neurologists, Other Relevant Specialities
Testing: Diagnostic testing offered in clinically affected patients, presymptomatic testing in patients at risk of developing HNPP (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 deletion of *PMP22*. This test will also detect the 1.5 Mb *PMP22* duplication associated with Charcot Marie Tooth disease type 1A (CMT1A).

Service Details:

Approximately 84% of HNPP patients have a deletion of *PMP22* (Nelis *et al*. 1996 Eur. J. Hum Genet. 4:25-33). Point mutations in *PMP22* have been identified in a small proportion of patients with the deletion. DNA can be sent to another centre for this testing, but the cost of the 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.