Hereditary Neuropathy with Liability to Pressure Palsies (HNPP) – OMIM 162500

Background

HNPP is caused by a mutation (often a deletion) in the gene *PMP22* (OMIM 601097) and follows a dominant inheritance pattern, so a child has a 50% chance of inheriting the disease from an affected parent. Charcot-Marie-Tooth Type 1A (CMT1A) is caused by a mutation in the same gene, and similar symptoms can lead to misdiagnosis. The incidence rate of HNPP in the UK is about 2 to 5 in 100,000 people having the condition, although it is thought that many more go undiagnosed or misdiagnosed. The majority of people with HNPP have mild symptoms, the main problem being pressure palsies – pressure on certain nerves causes tingling, numbness or sometimes muscle weakness. The main areas of the body affected tend to be legs, feet, elbows, wrists and hands. The condition evolves very slowly; the age of onset can vary from birth onwards but most commonly symptoms are noticed as a young adult. Although the quality of life for some individuals is affected, HNPP does not affect lifespan.

Recommended Clinical Referral Criteria

- Diagnostic cases and at-risk family members
- Autosomal dominant inheritance
- Recurrent focal compression neuropathies

Molecular Analysis

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

Test (Price available on request)	**TAT (calendar days)**
Dosage analysis (*PMP22)* (R77)	42
Predictive analysis	28

Contact Details
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www.medicalgenomicswales.co.ukAccredited 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 at least two identifiers i.e. name and date of birth

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/
EDDNAL
http://www.eddnal.com/
OMIM
http://www.omim.org/
Genetic Test Registry
http://www.ncbi.nlm.nih.gov/gtr/

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