TEST ID: PMP22
PMP22, PERIPHERAL NEUROPATHY, FISH

USEFUL FOR
Diagnosis of Charcot-Marie-Tooth type 1A or hereditary neuropathy with liability to pressure palsies

CLINICAL INFORMATION
This test is appropriate for individuals with clinical features suggestive of Charcot-Marie-Tooth type 1A (CMT1A) and hereditary neuropathy with liability to pressure palsies (HNPP).
CMT1A is a dominantly inherited disease caused by a duplication of the proximal short arm of chromosome 17, including the PMP22 gene. Clinical characteristics of CMT1A include progressive distal muscle weakness and atrophy, sensory loss, and slow nerve conduction velocity starting early in life.

SPECIMEN REQUIRED
Specimen Type
Blood
Container/Tube
Green top (sodium heparin)
Specimen Volume
6 mL

INTERPRETATION
- An interpretive report is provided.
- The presence of duplication of PMP22 confirms the diagnosis of Charcot-Marie-Tooth type 1A.
- The presence of heterozygous deletion of PMP22 confirms the diagnosis of hereditary neuropathy with liability to pressure palsies.

SUPPORTIVE DATA
FISH analysis was performed on a series of 12 fixed cell pellet samples previously identified with 17p12 deletions or duplications, and 25 normal blood and bone marrow control specimens. Deletions or duplications of the PMP22 gene region were verified and concordant in all 12 samples. The normal control samples were used to generate a normal cutoff for this assay.

MOBILE APPS FROM MAYO MEDICAL LABORATORIES
Lab Catalog for iPad and Lab Reference for iPhone and iPod Touch
Requires iOS 5.1+

REFERENCE VALUES
An interpretive report will be provided.

ANALYTIC TIME
7 days

CONTENT AND VALUES SUBJECT TO CHANGE. SEE THE MAYO MEDICAL LABORATORIES TEST CATALOG FOR CURRENT INFORMATION.
CLINICAL REFERENCE

