

# Phytanic acid

## General:

Phytanic acid is a saturated and branched-chain fatty acid, which is usually degraded by oxidation to 2, 6, 10, 14- tetramethylpentadecanoic acid. In REFSUM syndrome, it accumulates in serum and in organs. REFSUM's disease or heredopathia atactica polyneuritiformis (HAP), is an autosomalrecessive peroxisomal metabolic disorder (storage of phytanic acid as a result oxidation deficiency, whereby the 3, 7, 11, 15-tetramethyl hexadecanoic acid can not enter into  $\beta$ -oxidation). Manifestation in the first and second decade of life leads to symmetric polyneuropathy (especially legs), cerebellar ataxia, retinitis pigmentosa and hemeralopia, polyneuropathy, bone resorption, ichthyosis, albumin accumulation in CSF (with normal cell count).

Indication: REFSUM syndrome

Material: 1 ml serum

TAT: 10-14 days\*

Method: GCMS

Units: mg/l

Ref.- range: up to 5.00

For complete list of laboratory test offered at Freiburg Medical Laboratory, please visit <http://www.fml-dubai.com/parameter-listings/>