DISEASE:
Refsum disease

NAME:	Refsum disease
DESCRIPTION:	A metabolic disease characterized by anosmia, cataract, early-onset retinitis pigmentosa and possible neurological manifestations, including peripheral neuropathy and cerebellar ataxia. Other features can be deafness, ichthyosis, skeletal abnormalities, and cardiac arrhythmia. It is characterized biochemically by accumulation of phytanic acid in plasma and tissues.
ORPHACODE:	773
SYNONYMS:	Adult Refsum disease Classic Refsum disease HMSN 4 HMSN IV Hereditary motor and sensory neuropathy type 4 Hereditary motor and sensory neuropathy type IV Heredopathia atactica polyneuritiformis Phytanic-CoA hydroxylase deficiency

XREF(S):	Orphanet OMIM OMIM MeSH MedDRA ICD-10
ANALYTE(S):	PHYH PEX7
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RELATED CONTENT

Related Genetic Tests

- Ichthyosis (gene panel)
- Neuropathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- peroxisomal biogenesis factor 7
- phytanoyl-CoA 2-hydroxylase

Related Gene Panels

- Ichthyosis and erythroderma (98 genes) - KUL
- Neuropathy (148 genes) - IPG