DISEASE:
Polyneuropathy-hearing loss-ataxia-retinitis pigmentosa-cataract syndrome

NAME:	Polyneuropathy-hearing loss-ataxia-retinitis pigmentosa-cataract syndrome
DESCRIPTION:	This rare neurologic disease is a slowly-progressive Refsum-like disorder associating signs of peripheral neuropathy with late-onset hearing loss, cataract and pigmentary retinopathy that become evident during the third decade of life.
ORPHACODE:	171848
SYNONYMS:	PHARC syndrome Peripheral neuropathy, Fiskerstrand type Polyneuropathy-deafness-ataxia-retinitis pigmentosa-cataract syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ABHD12
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Charcot-Marie-Tooth (other than type 1A) (gene panel, IPN panel)
- Neuropathy (gene panel)

Related Laboratories

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

Related Analytes

- abhydrolase domain containing 12, lysophospholipase

Related Gene Panels

- Inherited Peripheral Neuropathies gene panel (139 genes) - KUL
- Neuropathy (148 genes) - IPG