DISEASE:
Fatty acid hydroxylase-associated neurodegeneration

NAME:	Fatty acid hydroxylase-associated neurodegeneration
DESCRIPTION:	Fatty acid hydroxylase-associated neurodegeneration (FAHN) is a very rare, autosomal recessive form of neurodegeneration with brain iron accumulation (NBIA) characterized by childhood-onset focal dystonia, progressive spastic paraplegia that progresses to tetra paresis, ataxia, dysarthria, intellectual decline, and oculomotor disturbances (optic atrophy), accompanied by iron deposition in the globus pallidus.
ORPHACODE:	329308
SYNONYMS:	FAHN
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	FA2H
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RELATED CONTENT

Related Genetic Tests

- Neurodegeneration with Brain Iron Accumulation (gene panel)
- Spastic Paraplegia (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Gent

Related Analytes

- fatty acid 2-hydroxylase

Related Gene Panels

- Neurodegeneration with Brain Iron Accumulation (NBIA) - UGent
- Spastic Paraplegia (89 genes) - IPG

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