DISEASE:
Familial dysautonomia

NAME:	Familial dysautonomia
DESCRIPTION:	A rare hereditary sensory and autonomic neuropathy characterized by decreased pain and temperature perception, absent deep tendon reflexes, proprioceptive ataxia, afferent baroreflex failure and progressive optic neuropathy.
ORPHACODE:	1764
SYNONYMS:	HSAN3 Hereditary sensory and autonomic neuropathy type 3 Hereditary sensory and autonomic neuropathy type III Riley-Day syndrome
XREF(S):	Orphanet MedDRA OMIM ICD-10 MeSH
ANALYTE(S):	ELP1
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RELATED CONTENT

Related Genetic Tests

- Dysautonomia, familial (FD) (hot spot mutation - c.2204+6T>C)
- Jewish mutation panel (Tay Sachs, Fanconi, Dysautonomia, Canavan) (4 genes; 7 hot spot mutations)
- Neuropathy (gene panel)

Related Laboratories

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

Related Analytes

- elongator complex protein 1

Related Gene Panels

- Hot spot mutation among Jewish (4 genes, 7 mutations) - UZA
- Neuropathy (148 genes) - IPG