DISEASE:
Hereditary sensory and autonomic neuropathy type 6

NAME:	Hereditary sensory and autonomic neuropathy type 6
DESCRIPTION:	A rare hereditary sensory and autonomic neuropathy characterized by hypotonia in infancy, variable psychomotor retardation, markedly impaired pain sensitivity with poorly healing distal ulcerations and painless fractures leading to joint deformities and amputation of fingers and toes, altered deep tendon reflexes, and dysautonomic symptoms including hypohidrosis and heat intolerance, chronic diarrhea, pupillary abnormalities, or urinary incontinence. Sensorineural hearing loss has also been reported. The severity of the disease is highly variable, with severe cases being potentially lethal in infancy.
ORPHACODE:	314381
SYNONYMS:	Familial dysautonomia with contractures HSAN6 Hereditary sensory and autonomic neuropathy type VI
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	DST
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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

- dystonin

Related Gene Panels

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