DISEASE:
Hereditary sensory and autonomic neuropathy type 8

NAME:	Hereditary sensory and autonomic neuropathy type 8
DESCRIPTION:	A rare autosomal recessive hereditary sensory and autonomic neuropathy characterized by congenital impaired sensation of acute or inflammatory pain in combination with an inability to identify noxious heat or cold, leading to numerous painless mutilating lesions and injuries. Further manifestations are absence of corneal reflexes resulting in corneal scarring, reduced sweating and tearing, and recurrent skin infections. Large-fiber sensory modalities such as light touch, vibration, and proprioception are normal.
ORPHACODE:	478664
SYNONYMS:	HSAN8 Hereditary sensory and autonomic neuropathy type VIII
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	PRDM12
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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

- PR/SET domain 12

Related Gene Panels

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