DISEASE:
Hereditary sensory and autonomic neuropathy due to TECPR2 mutation

NAME:	Hereditary sensory and autonomic neuropathy due to TECPR2 mutation
DESCRIPTION:	A rare genetic peripheral neuropathy characterized by early hypotonia evolving to spastic paraparesis, areflexia, decreased pain and temperature sensitivity, autonomic neuropathy, gastroesophageal reflux disease, recurrent pneumonia and respiratory problems. Patients also have intellectual disability and dysmorphic features, including mild brachycephalic microcephaly, short broad neck, low anterior hairline and coarse face.
ORPHACODE:	320385
SYNONYMS:	Autosomal recessive spastic paraplegia type 49 HSAN due to TECPR2 mutation SPG49
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	TECPR2
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- Spastic Paraplegia (89 genes) - IPG

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