Opis choroby *

Definicja

Hereditary sensory and autonomic neuropathy due to TECPR2 mutation is 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.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive spastic paraplegia type 49
SPG49
HSAN due to TECPR2 mutation
SPG49

Kod ORPHA
320385

Kod OMIM
615031

Kod ICD10
G11.4

Kod ICD11
8C21.Y

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*Źródło

orphanet