Opis choroby *

Definicja

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.

Dane

Klasyfikacja
Choroba

Synonimy
HSAN8
Dziedziczna neuropatia czuciowa i autonomiczna typu VIII
Dziedziczna neuropatia czuciowa i autonomiczna typu 8
HSAN8
Zespół CIP i hipohydrozy
Hereditary sensory and autonomic neuropathy type VIII

Kod ORPHA
478664

Kod OMIM
616488

Kod ICD10
G60.8

Kod ICD11
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*Źródło

orphanet