Opis choroby *

Definicja

A rare, autosomal dominant neurological disorder due to truncation mutations of the prion protein gene PRNP (20p13) leading to deposition of prion protein amyloid. Onset is usually in the fourth decade of life and reported clinical manifestations include diarrhea, nausea, autonomic failure (areflexia, weakness), neurogenic bladder and urinary infections.

Dane

Klasyfikacja
Choroba

Synonimy
Chronic diarrhea with HSAN
Przewlekła biegunka z HSAN
Chronic diarrhea with hereditary sensory and autonomic neuropathy
Prion protein systemic amyloidosis

Kod ORPHA
397606

Kod OMIM
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Kod ICD10
G60.8

Kod ICD11
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*Źródło

orphanet