Opis choroby *

Definicja

A rare lysosomal storage disease characterized clinically by severe global development delay due to neuronal dysmyelination, hypotonia which gradually progresses to spasticity during childhood, speech deficits, progressive visual impairment (due to corneal clouding, retinal degeneration and optic atrophy), achlorhydria, with increased gastrin secretion and iron deficiency anemia, and kidney disease and failure, all in the absence of dysmorphic features.

Dane

Klasyfikacja
Choroba

Kod ORPHA
578

Kod OMIM
252650

Kod ICD10
E75.1

Kod ICD11
5C56.0Y

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*Źródło

orphanet