Opis choroby *

Definicja

A rare, autosomal recessive, acid sphingomyelinase deficiency characterized clinically by onset in infancy or early childhood with failure to thrive, hepatosplenomegaly, interstitial lung disease and rapidly progressive neurodegenerative disorders.

Dane

Klasyfikacja
Choroba

Synonimy
Infantile neurovisceral ASMD
Niemowlęcy niedobór kwaśnej sfingomielinazy nerwowo-trzewnej
NPD-A
Niemann-Pick disease type A

Kod ORPHA
77292

Kod OMIM
257200

Kod ICD10
E75.2

Kod ICD11
5C56.0Y

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*Źródło

orphanet