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

Kod ORPHA	**Kod OMIM**	**Kod ICD10**
77292	257200	E75.2

Kod ICD11
5C56.0Y

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*Źródło

orphanet