

Opis choroby *

Definicja

Free sialic acid storage disease (free SASD), is a group of lysosomal storage diseases characterized by a spectrum of clinical manifestations including neurological and developmental disorders with severity ranging from the milder phenotype, Salla disease (SD), to the most severe phenotype, infantile free sialic acid storage disease (ISSD).

Dane

Klasyfikacja

Choroba

Kod ORPHA

834

Kod OMIM

604369

Kod ICD10

E77.8

Kod ICD11

5C56.4

*Źródło

orphanet