

Opis choroby *

Definicja

A very rare lysosomal storage disease that is clinically and pathologically heterogeneous and is characterized by deficient NAGA activity.

Dane

Klasyfikacja

Choroba

Synonimy

NAGA deficiency
Choroba Schindlera
Niedobór NAGA
Schindler disease

Kod ORPHA

3137

Kod OMIM

609242

Kod ICD10

E77.1

Kod ICD11

5C56.21

*Źródło

orphanet