

## 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

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#### \*Źródło

orphanet