

## Opis choroby \*

### Definicja

Gaucher disease type 2 is the acute neurological form of Gaucher disease (GD; see this term). It is characterized by early-onset and severe neurological involvement of the brainstem, associated with an organomegaly and generally leading to death before the age of 2.

### Dane

#### Klasyfikacja

Podtyp kliniczny

#### Synonimy

Acute neuronopathic Gaucher disease

Dziecięca mózgowa choroba Gauchera

Ostra neuropatyczna choroba Gauchera

Infantile cerebral Gaucher disease

#### Kod ORPHA

77260

#### Kod OMIM

230900

#### Kod ICD10

E75.2

#### Kod ICD11

5C56.0Y

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#### \*Źródło

orphanet