

## Opis choroby \*

### Definicja

Galactosialidosis is a lysosomal storage disease characterized by coarse facial features, macular "cherry red spot", and dysostosis multiplex. Clinical presentation can be heterogenous ranging from a severe, early-onset, rapidly progressive infantile form to late onset, slowly progressive juvenile/adult form.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Goldberg syndrome

Niedobór neuraminidazy z niedoborem beta-galaktozydazy

Zespół Goldberga

Neuraminidase deficiency with beta-galactosidase deficiency

#### Kod ORPHA

351

#### Kod OMIM

256540

#### Kod ICD10

E77.1

#### Kod ICD11

5C56.21

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#### \*Źródło

orphanet