

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 | Synonimy |
|--------------|---|
| Choroba | Goldberg syndrome |
| | Niedobór neuraminidazy z niedoborem beta-galaktozydazy |
| | Zespół Goldberga |
| | Neuraminidase deficiency with beta-galactosidase deficiency |

| Kod ORPHA | Kod OMIM | Kod ICD10 |
|-----------|----------|-----------|
| 351 | 256540 | E77.1 |

| Kod ICD11 |
|-----------|
| 5C56.21 |

*Źródło

orphanet