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