Opis choroby *

Definicja

GM1 gangliosidosis is a rare lysosomal storage disorder characterized biochemically by deficient beta-galactosidase activity and clinically by a wide range of variable neurovisceral, ophthalmological and dysmorphic features.

Dane

Klasyfikacja
Choroba

Synonimy
Beta-galactosidase-1 deficiency
Choroba Landinga
Niedobór beta-galaktozydazy-1
Niedobór GLB1
GLB1 deficiency
Landing disease

Kod ORPHA
354

Kod OMIM
230650

Kod ICD10
E75.1

Kod ICD11
5C56.00

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*Źródło

orphanet