

# Niedobór syntazy GM3

Kod Orpha: 370933 Kod OMIM: 609056

## Opis choroby \*

### Definicja

GM3 synthase deficiency is a rare congenital disorder of glycosylation due to impaired synthesis of complex ganglioside species initially characterized by irritability, poor feeding, failure to thrive and early-onset refractory epilepsy, followed by postnatal growth impairment, severe developmental delay or developmental regression, profound intellectual disability, deafness and abnormalities of skin pigmentation (mostly freckle-like hyperpigmented and depigmented macules). Visual impairment due to cortical atrophy (visible on magnetic resonance imaging), choreoathetosis and hypotonic tetraparesis usually appear gradually. Dysmorphic facial features may be associated.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

ST3GAL5-CDG

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#### Kod ORPHA

370933

#### Kod OMIM

609056

#### Kod ICD10

E77.8

#### Kod ICD11

5C54.Y

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#### [\\*Źródło](#)

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## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

Dostępna na stronie [www.orphanet.pl](http://www.orphanet.pl)