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
ST3GAL5-CDG

Kod ORPHA
370933

Kod OMIM
609056

Kod ICD10
E77.8

Kod ICD11
5C54.Y

---

*Źródło

orphanet