## Opis choroby *

Definicja

GM2 gangliosidosis, AB variant is an extremely rare, severe genetic disorder characterized by progressive neurological decline due to ganglioside activator deficiency.

Dane

**Klasyfikacja**
Choroba

Synonimy
Hexosaminidase activator deficiency
Niedobór aktywatora heksozoaminidazy

**Kod ORPHA**
309246

**Kod OMIM**
272750

**Kod ICD10**
E75.0

**Kod ICD11**
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*Źródło

orphanet