Opis choroby *

Definicja

A rare, severe form of mucolipidosis characterized by growth retardation, skeletal abnormalities (dysostosis multiplex, craniosynostosis, contractures of the joints and osteopenia), facial dysmorphism, stiff skin, obstructive airway, cardiomegaly and severe global developmental delay.

Dane

Klasyfikacja
Choroba

Synonimy
I-cell disease
Niedobór N-acetyloglukozamino-1-fosfotransferazy
Choroba "I-cell"
Mucolipidosis type II alpha/beta
N-acetylglucosamine 1-phosphotransferase deficiency

Kod ORPHA
576

Kod OMIM
252500

Kod ICD10
E77.0

Kod ICD11
5C56.20

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*Źródło

orphanet