## 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