

Opis choroby *

Definicja

A rare, severe form of mucopolysaccharidosis 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"

Mucopolysaccharidosis type II alpha/beta

N-acetylglucosamine 1-phosphotransferase
deficiency

Kod ORPHA

576

Kod OMIM

252500

Kod ICD10

E77.0

Kod ICD11

5C56.20

*Źródło

orphanet