

Mukolipidoza typu III gamma

Kod Orpha: 423470 Kod OMIM: 252605

Opis choroby *

Definicja

Mucopolipidosis type III gamma (ML 3 gamma) is a very rare lysosomal disease, that has most often been observed in the Middle East, characterized by a progressive slowing of the growth rate in early childhood; stiffness and pain in shoulders, hips, and finger joints; a gradual, mild coarsening of facial features; and by a slower progression, milder clinical course and longer life expectancy than that seen in mucopolipidosis type II and mucopolipidosis type III alpha/beta. Cognitive function is normal or only slightly impaired and retinitis pigmentosa has been reported in a few patients. Many survive into early adulthood, but ultimately succumb to cardiorespiratory insufficiency.

Dane

Klasyfikacja

Podtyp kliniczny

Synonimy

ML 3 gamma
ML 3 gamma
ML III gamma
Mukolipidoza typu 3 gamma
ML III gamma
Mucopolipidosis type 3 gamma

Kod ORPHA

423470

Kod OMIM

252605

Kod ICD10

E77.0

Kod ICD11

5C56.20

[*Źródło](#)

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Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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