Opis choroby *

Definicja

Mucolipidosis 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 mucolipidosis type II and mucolipidosis 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
Mucolipidosis type 3 gamma

Kod ORPHA
423470

Kod OMIM
252605

Kod ICD10
E77.0

Kod ICD11
5C56.20

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*Źródło

orphanet