

Opis choroby *

Definicja

Mucopolipidosis III alpha/beta (MLIII alpha/beta) is a lysosomal disorder characterized by progressive slowing of the growth rate from early childhood, stiffness and pain in joints, gradual coarsening of facial features, moderate developmental delay and mild intellectual disability in most patients.

Dane

Klasyfikacja	Synonimy
Podtyp kliniczny	ML 3 alpha/beta ML 3 alfa/beta ML III alfa/beta Mukolipidoza typu 3 alfa/beta ML III alpha/beta Mucopolipidosis type 3 alpha/beta

Kod ORPHA	Kod OMIM	Kod ICD10
423461	252600	E77.0

Kod ICD11
5C56.20

*Źródło

orphanet