Opis choroby *

Definicja

A rare, congenital disorder of glycosylation-related bone disorder characterized by hypotonia, severe developmental delay, intellectual disability, seizures, increased serum alkaline phosphatase, short distal phalanges with hypoplastic nails, and dysmorphic facial features. In some cases, cleft palate, megacolon, anorectal malformations, and congenital heart defects have been reported.

Dane

Klasyfikacja
Choroba

Synonimy
Mabry syndrome
HPMR
Zespół Mabry'ego

Kod ORPHA
247262

Kod OMIM
616809

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet