Opis choroby *

Definicja

A rare genetic disease characterized by mild intellectual disability, osteoporosis, delayed bone age, macrocephaly with wormian bones and frontal bossing, anomalies of fingers, nails, and teeth, thoracic deformities, hyperextensibility of joints, as well as congenital amaurosis and paraplegia. There have been no further descriptions in the literature since 1981.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Heide syndrome
Zespół Heidego

Kod ORPHA
2787

Kod OMIM
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Kod ICD10
Q87.5

Kod ICD11
LD24.KY

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*Źródło

orphanet