Opis choroby *

Definicja

A rare primary bone dysplasia characterized by megalocornea, multiple skeletal anomalies, characteristic facial dysmorphism (wide fontanels, prominent forehead, hypertelorism, prominent eyes, full cheeks and micrognathia) and developmental delay.

Dane

Klasyfikacja
Choroba

Synonimy
Ter Haar syndrome
Zespół Ter Haara

Kod ORPHA
137834

Kod OMIM
249420

Kod ICD10
Q87.8

Kod ICD11
LD25.1

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*Źródło

orphanet