Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by the association of developmental delay and mild chondrodysplasia with short stature and abnormal growth plate morphology. Dysmorphic facial features are variable and may include hypertelorism, upslanting palpebral fissures, broad nose with broad nasal tip, and low-set, cup-shaped ears, among others. Autism spectrum disorder and neurologic abnormalities have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
580940

Kod OMIM
617982

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet