Opis choroby *

Definicja

A very rare multiple congenital anomalies syndrome characterized by short stature, facial dysmorphism (elongated face, hypertelorism, broad and high nasal bridge, mild epicanthus, posteriorly angulated ears, narrow and high-arched palate), skeletal anomalies (mesomelic brachymelia, short broad hands, prominent finger pads, short stubby thumbs, hyperextensibility of small joints, small feet), hypernasality and normal intelligence. Delayed bone age has also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
3424

Kod OMIM
600736

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet