Opis choroby *

Definicja

A multiple congenital developmental anomalies syndrome characterized by arachnodactyly of fingers and toes associated with craniofacial dysmorphism (including abnormal cranial ossification, frontal bossing, flat calvaria, shallow deformed orbits resulting in exophtalmos, midface hypoplasia and micrognathia), feeding difficulties in infancy, infantile muscular hypotonia, and developmental delay leading to intellectual disability.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Kosztolanyi syndrome
Zespół Kosztolanyi

Kod ORPHA
1129

Kod OMIM
-

Kod ICD10
Q87.8

Kod ICD11
-

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*Źródło

orphanet