

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 exophthalmos, midface hypoplasia and micrognathia), feeding difficulties in infancy, infantile muscular hypotonia, and developmental delay leading to intellectual disability.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Kosztolanyi syndrome
	Zespół Kosztolanyi

Kod ORPHA	Kod OMIM	Kod ICD10
1129	-	Q87.8

Kod ICD11
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*Źródło

orphanet