Opis choroby *

Definicja

A rare multiple congenital anomalies syndrome characterized by variable skeletal abnormalities (including craniostenosis, pectus carinatum, short sternum, joint hyperextensibility, and anbnormal vertebrae), cutis laxa with excessive skin folds around the cheek, chin and neck, ambiguous genitalia with a micropenis and perineal hypospadia, an umbilical hernia, intellectual disability, premature aged appearance, and cardiac enlargement involving either the ventricles or atria. Facial dysmorphism is variable and can include multiple hair whorls, ptsosis, high and broad nasal root, low set ears and small chin. Enamel hypocalcification, abnormal modelling of tubular bones, and reduced cutis laxa may become apparent later on.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
3134

Kod OMIM
312830

Kod ICD10
Q82.8

Kod ICD11
LD28.2

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*Źródło

orphanet