Opis choroby *

Definicja

A rare multiple congenital anomalies syndrome characterized by the association of camptodactyly, multiple eye defects (fibrosis of the medial rectus muscle, severe myopia, ptosis and exophthalmos), scoliosis, flexion contractures and facial anomalies (arched eyebrows, facial asymmetry with an abnormal skull shape, a prominent nose, small mouth, low-set and dysplastic ears, and a low nuchal hairline).

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Rozin camptodactyly syndrome
Zespół Rozina, Hertza i Goodmana
Zespół Rozina - kamptodaktylia

Kod ORPHA
1323

Kod OMIM
602612

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet