

Opis choroby *

Definicja

A rare orofacial clefting syndrome characterized by the association of Pierre Robin sequence (retrognathia, cleft palate and glossoptosis) with facial dysmorphism (high forehead with frontal bossing) and digital anomalies (tapering fingers, hyperconvex nails, clinodactyly of the fifth fingers and short distal phalanges, finger-like thumbs and easily subluxated first metacarpophalangeal joints). Growth and mental development were normal.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Chitayat-Meunier-Hodgkinson syndrome

Sekwencja Pierre'a Robina - anomalia twarzowo-palcowa

Zespół Chitayata, Meuniera i Hodgkinsona

Pierre Robin sequence-faciodigital anomaly syndrome

Kod ORPHA

2888

Kod OMIM

311895

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet