Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by short stature and particularly pronounced shortening of the third to fifth metacarpals and metatarsals, congenital anodontia, sparse hair, dyspigmentation of the skin, hypoplastic nipples and underdeveloped external genitals in females, and multiple ocular abnormalities (such as distichiasis, strabismus, nystagmus, lenticular opacities, and severe myopia, among others). Dysmorphic craniofacial features include brachycephaly, downslanting palpebral fissures, broad nasal root, low-set ears, and small maxilla and prominent mandible. There have been no further descriptions in the literature since 1968.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
2713

Kod OMIM
211370

Kod ICD10
Q87.5

Kod ICD11
LD27.0Y

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*Źródło

orphanet