

Opis choroby *

Definicja

Craniofaciofrontodigital syndrome is a rare multiple congenital anomalies syndrome characterized by mild intellectual disability, short stature, cardiac anomalies, mild dysmorphic features (macrocephaly, prominent forehead, hypertelorism, exophthalmos), cutis laxa, joint hyperlaxity, wrinkled palms and soles and skeletal anomalies (sella turcica, wide ribs and small vertebral bodies).

Dane

Klasyfikacja

Choroba

Synonimy

Cantu craniofaciofrontodigital syndrome
Zespół czaszkowo-twarzowo-czołowo-palcowy
Cantu

Kod ORPHA

363705

Kod OMIM

114620

Kod ICD10

Q87.0

Kod ICD11

-

*Źródło

orphanet