

Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by the association of intellectual deficit, facial dysmorphism (a highly arched palate, pointed chin, and small mouth, hypotelorism, a long nose and large protruding ears), arachnodactyly, hypogenitalism (undescended testes and hypospadias) and failure to thrive.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Cranio-facio-digito-genital syndrome

Zespół czaszkowo-twarzowo-palcowo-płciowy

Kod ORPHA

2115

Kod OMIM

601095

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet