Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by variable developmental delay and intellectual disability, overweight or obesity, behavioral abnormalities (including hyperactivity, aggressive behavior, anxiety, mood disorder, or autistic features), and facial dysmorphism (such as high forehead, full eyebrows and/or synophrys, upturned nose, and fleshy ears, among others). Additional reported manifestations are hypotonia, ocular anomalies, anomalies of the fingers and toes, joint hypermobility, or abnormal pigmentation. Brain imaging may show mild nonspecific abnormalities.

Dane

Klasyfikacja
Choroba

Synonimy
Chung-Jansen syndrome
Zespół Chung i Jansen
DIDOD

Kod ORPHA
589905

Kod OMIM
617991

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet