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 Synonimy

Choroba Chung-Jansen syndrome

Zespół Chung i Jansen

DIDOD

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 589905
 617991
 Q87.8

Kod ICD11

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*Źródło

orphanet