Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by global developmental delay, intellectual disability, short stature, skeletal abnormalities (such as brachydactyly and vertebral anomalies), obesity, cardiac, respiratory, and genitourinary anomalies, and dysmorphic facial features (including coarse facies, thick eyebrows, synophrys, hypertelorism, short, upturned nose, and long philtrum). Additional reported manifestations are microcephaly, hearing impairment, cataract, and gastroesophageal reflux.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
CHOPS syndrome
Zespół CHOPS

Kod ORPHA
444077

Kod OMIM
616368

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet