Opis choroby *

Definicja

A rare genetic, multiple congenital anomalies syndrome characterized by short stature, hand brachydactyly with hypoplastic distal phalanges, global development delay, intellectual disability, and more variably seizures, obesity, and craniofacial dysmorphism that includes microcephaly, high forehead, flat face, hypertelorism, deep set eyes, flat nasal bridge, averted nostrils, long philtrum, thin lip vermilion, and short neck.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
SBIDDS
SBIDDS

Kod ORPHA
464288

Kod OMIM
617157

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet