

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	Synonimy
Zespół wad wrodzonych	SBIDDS
	SBIDDS

Kod ORPHA	Kod OMIM	Kod ICD10
464288	617157	Q87.8

Kod ICD11
-

*Źródło

orphanet