Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by global developmental delay, mild intellectual disability, seizures, obesity, and dysmorphic facial features (including large, bulbous nose, prominent philtrum, wide mouth). Additional reported features are bilateral pes planus, scoliosis, and spina bifida occulta. Brain MRI may show mild ventricular dilatation.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Intellectual disability-epilepsy-bulbous nose syndrome
Niepełnosprawność intelektualna - padaczka - kartoflowaty nos

Kod ORPHA
2139

Kod OMIM
-

Kod ICD10
Q87.8

Kod ICD11
-

---

*Źródło

orphanet