

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

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Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet