

Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome with variable intellectual disability characterized by abnormal head shape/metopic ridging and facial dysmorphism (which may include arched eyebrows, ptosis, downslanting palpebral fissures, epicanthal folds, and short upturned nose). Many patients present variable global developmental delay and/or autism spectrum disorder. Additional reported features are cardiac, skeletal, or urogenital anomalies. Brain imaging may show agenesis of the corpus callosum.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

502430

Kod OMIM

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Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet