Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by global developmental delay, variable degrees of intellectual disability, and facial dysmorphism (including high nasal bridge, deep-set eyes, and wide mouth), often associated with feeding difficulties and/or gastroesophageal reflux. Additional reported manifestations are seizures, hypotonia, autistic features, and joint laxity. Brain imaging may show non-specific features (such as cerebral atrophy).

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
502434

Kod OMIM
617635

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet