

## 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