

## Opis choroby \*

### Definicja

A rare genetic neurological disorder characterized by infantile to childhood onset of global developmental delay, hypotonia, seizures, growth delay, and intellectual disability. Additional variable features include strabismus, cortical visual impairment, nystagmus, movement disorder (such as dystonia, ataxia, or chorea), or mild dysmorphic features, among others.

### Dane

### Klasyfikacja

Zespół wad wrodzonych

#### Kod ORPHA

488613

#### Kod OMIM

616973

#### Kod ICD10

F84.8

#### Kod ICD11

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### \*Źródło

orphanet