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