Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by variable degrees of developmental delay and intellectual disability with poor or absent speech, hypotonia, hypoplastic or absent corpus callosum, and facial dysmorphism (such as long face, frontal bossing, hypertelorism, downslanting palpebral fissures, and tented upper lip). Additional reported features include microcephaly, seizures, gait ataxia, scoliosis, and syndactyly of fingers, among others.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
457284

Kod OMIM
616362

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet