Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by global developmental delay, intellectual disability, growth retardation, hypotonia, cerebellar symptoms such as ataxia, spondyloepiphyseal dysplasia, and dysmorphic craniofacial features (including microcephaly, dolichocephaly, prominent ears, epicanthus, broad nasal bridge, long and flat philtrum, or small mouth). Additional reported manifestations are epilepsy, retinitis pigmentosa, and urogenital abnormalities, among others. Brain imaging may show cerebellar hypoplasia.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
459070

Kod OMIM
300998

Kod ICD10
Q87.0

Kod ICD11
LD90

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*Źródło

orphanet