Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by profound intellectual disability, hypotonia, coarse facial features, strabismus and impaired visual fixation, hypermobility of interphalangeal joints, contractures in the elbow joints, and pes planovalgus. Seizures and episodes of aggressive behavior during sleep have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
436141

Kod OMIM
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Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet