Opis choroby *

Definicja

A rare, genetic, syndromic intellectual disability disorder characterized by severe psychomotor development delay (without development of primary motor abilities and speech) and sever intellectual disability, associated with marfanoid habitus, joint laxity, bilateral hip luxation, hypotonia, scoliosis, and characteristic facial dysmorphism (i.e. high nasal bridge, sharp nose, short philtrum, large mouth, full lips and maxillary hypoplasia). There have been no further description in the literature since 1994.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
2058

Kod OMIM
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Kod ICD10
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Kod ICD11
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*Źródło

orphanet