

Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by intellectual disability, psychomotor retardation, flat face and some features resembling Marfan syndrome, such as tall stature, dolichostenomelia, arm span larger than height, arachnodactyly of hands and feet, little subcutaneous fat, and muscle hypotonia. There have been no further descriptions in the literature since 1984.

Dane

Klasyfikacja

Synonimy

Zespół wad wrodzonych Fragoso-Cantú syndrome

Kod ORPHA

2463

Kod OMIM

248770

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet