Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by trigonobrachycephaly, facial dysmorphism (including narrow forehead, upward-slanting palpebral fissures, bulbous nose with slightly bifid tip, macrostomia with thin upper lip, micrognathia), and various acral anomalies, such as broad thumbs, large toes, bulbous fingertips with short nails, joint laxity of the hands and fifth finger clinodactyly. Short stature, hypotonia and severe psychomotor delay are also associated. There have been no further descriptions in the literature since 1991.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
3368

Kod OMIM
275595

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet