Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by global developmental delay, intellectual disability, growth retardation, hearing impairment, characteristic facial dysmorphology (including prominent supraorbital ridges, downslanting palpebral fissures, deep-set eyes, long face, sagging cheeks, anteverted nares, and pointed chin), generalized hypotonia, joint hypermobility, gluteal crease with sacral caudal remnant and sacral dimple, and variable neurological features. Various ophthalmic, cutaneous, musculoskeletal, gastrointestinal, and cardiovascular anomalies have also been described.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
480907

Kod OMIM
300966

Kod ICD10
Q87.0

Kod ICD11
LD90

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*Źródło

orphanet