Opis choroby *

Definicja

Camptodactyly syndrome, Guadalajara type 3 is a rare, genetic bone development disorder characterized by hand camptodactyly associated with facial dysmorphism (flat face, hypertelorism, telecanthus, symblepharon, simplified ears, retrognathia) and neck anomalies (short neck with stricking pterygia, muscle sclerosis). Additional features include spinal defects (e.g. cervical and dorso-lumbar spina bifida occulta), congenital shortness of the sternocleidomastoid muscle, flexed wrists and thin hands and feet. Brain structural anomalies, multiple nevi, micropenis and mild intellectual disability are also observed. Imaging reveals increased bone traveculae, cortical thickening of long bones and delayed bone age.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
488434

Kod OMIM
611929

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet