

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 striking 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 trabeculae, 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