

Opis choroby *

Definicja

Cerebrofacioarticular syndrome is a rare multiple congenital anomalies syndrome characterized by mild to severe intellectual disability, a distinctive facial gestalt (blepharophimosis, maxillary hypoplasia, telecanthus, microtia and atresia of the external auditory meatus) as well as skeletal and articular abnormalities (e.g. camptodactyly of the fingers, cutaneous syndactyly, talipes equinovarus, flexion contractures of the proximal interphalangeal joints, hip or elbow subluxation, joint laxity). Affected individuals also present neonatal hypotonia, variable respiratory manifestations, chronic feeding difficulties and gray matter heterotopia.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Van Maldergem syndrome
	Zespół Van Maldergema

Kod ORPHA	Kod OMIM	Kod ICD10
314679	615546	Q87.8

Kod ICD11

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*Źródło

orphanet