Opis choroby *

Definicja

A type of arthrogryposis characterized by congenital cleft palate, microcephaly, craniostenosis and arthrogryposis (limitation of extension of elbows, flexed adducted thumbs, camptodactyly and clubfeet). Additional features include facial dysmorphism ('myopathic' stiff face, antimongoloid slanting, external ophthalmoplegia, telecanthus, low-set large malrotated ears, open mouth, mierogenia and high arched palate). Velopharyngeal insufficiency with difficulties in swallowing, increased secretion of the nose and throat, prominent occiput, generalized muscular hypotonia with mild cyanosis and no spontaneous movements, seizures, torticollis, areflexia, intellectual disability, hypertrichosis of the lower extremities, and scleredema (in the first days of life; see this term) are also observed. The disease often leads to early death. Transmission is autosomal recessive. No new cases have been described since 1983.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
2952

Kod OMIM
201550

Kod ICD10
Q74.8

Kod ICD11
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*Źródło

orphanet