Opis choroby *

Definicja

A developmental anomaly characterized at birth by the presence of right-sided aortic arch, craniofacial dysmorphism (microcephaly, asymmetric, facial bones, broad forehead, borderline hypertelorism, nasal septum deviation, large nasal cavity, large, posteriorly rotated ears, and microstomia with downturned corners), and intellectual disability. These features were observed in 4 members of one family, involving 2 successive generations, suggesting an autosomal dominant mode of transmission. There have been no further descriptions in the literature since 1968.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
1110

Kod OMIM
107500

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet