Aortic arch anomaly-facial dysmorphism-intellectual disability syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Aortic arch anomaly-facial dysmorphism-intellectual disability syndrome. ORPHA:1110

Aortic arch anomaly-peculiar facies-intellectual disability syndrome is 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.