8p23.1 duplication syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 8p23.1 duplication syndrome. ORPHA:251076

8p23.1 duplication syndrome is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the short arm of chromosome 8, with a highly variable phenotype, principally characterized by mild to moderate developmental delay, intellectual disability, mild facial dysmorphism (incl. prominent forehead, arched eyebrows, broad nasal bridge, upturned nares, cleft lip and/or palate) and congenital cardiac anomalies (e.g., atrioventricular septal defect). Other reported features include macrocephaly, behavioral abnormalities (e.g., attention deficit disorder), seizures, hypotonia and ocular and digital anomalies (poly/syndactyly).