Distal 22q11.2 microduplication syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal 22q11.2 microduplication syndrome. ORPHA:261337

Distal 22q11.2 microduplication syndrome is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the long arm of chromosome 22, with a highly variable phenotype principally characterized by developmental delay, intellectual disability, hypotonia, growth retardation, velopharyngeal insufficiency, mild craniofacial dysmorphism (microcephaly, tall/broad forehead, small downslating palpebral fissures, hooded eyelids, flat nasal bridge, low posterior hairline) and digital anomalies. Congenital heart malformations, visual and hearing impairment, urogenital abnormalities, behavourial problems and seizures have also been reported.