Distal monosomy 3p

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal monosomy 3p. ORPHA:1620

Distal monosomy 3p is a rare chromosomal anomaly syndrome, resulting from a partial deletion of the short arm of chromosome 3, with a highly variable phenotype typically characterized by pre- and post-natal growth retardation, intellectual disability, developmental delay and craniofacial dysmorphism (microcephaly, trigonocephaly, downslanting palpebral fissures, telecanthus, ptosis, micrognathia). Postaxial polydactyly, hypotonia, renal anomalies and congenital heart defects (e.g. atrioventricular septal defect) may be associated.