Trisomy 9p

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Trisomy 9p. ORPHA:236

Trisomy 9p is a rare chromosomal anomaly syndrome, resulting from a partial or complete trisomy of the short arm of chromosome 9, with a wide phenotypic variablility, typically characterized by intellectual disability, craniofacial dysmorphism (e.g. microcephaly, large anterior fontanel, hypertelorism, strabismus, downslanting palpebral fissures, malformed, low-set, protruding ears, bulbous nose, macrostomia, down-turned corners of mouth, micrognathia), digital anomalies (brachydactyly and clinodactyly), and short stature. Less frequently patients present with cardiopathy and renal, skeletal, and central nervous system malformations.