Distal monosomy 13q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal monosomy 13q. ORPHA:1590

Distal monosomy 13q is a rare chromosomal anomaly syndrome, resulting from a partial deletion of the long arm of chromosome 13, with a highly variable phenotype typically characterized by varying degrees of intellectual disability and developmental delay, as well as CNS malformations (e.g. holoprosencephaly, anencephaly, ventriculomegaly, Dandy-Walker malformation), ocular abnormalities (e.g. hypertelorism, microphthalmia, strabismus, aniridia, retinal dysplasia) and craniofacial dysmorphism (microcephaly, trigonocephaly, large and malformed ears, broad prominent nasal bridge, micrognathia). Cardiac, genitourinary, gastrointestinal and skeletal manifestations have also been reported.