Distal monosomy 7q36

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal monosomy 7q36. ORPHA:1636

Distal monosomy 7q36 is a rare chromosomal anomaly syndrome, resulting from a partial deletion of the long arm of chromosome 7, with a highly variable phenotype typically characterized by holoprosencephaly, growth restriction, developmental delay, facial dysmorphism (facial clefts, prominent forehead, hypertelorism, low-set ears, flat and broad nasal bridge, large mouth), abnormal fingers and palm or sole creases, ocular abnormalities, and other congenital malformations (incl. genital anomalies and caudal deficiency sequence). Cardiopathies have been occasionally reported.