Distal trisomy 7p

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal trisomy 7p. ORPHA:96074

Distal trisomy 7p is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the short arm of chromosome 7, with highly variable phenotype typically characterized by severe to profound psychomotor delay, intellectual disability, dysmorphic features (incl. dolichocephaly, microbrachycephaly, high and/or broad forehead, large anterior fontanel, hypertelorism, downslanting palpebral fissures, low-set, dysplastic ears, low, broad and prominent nasal bridge, abnormal palate, micro-/retrognathia), and hypotonia. Cardiovascular, gastrointestinal, skeletal and urogenital anomalies have commonly been reported.