Distal trisomy 1p36

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal trisomy 1p36. ORPHA:96069

Distal trisomy 1p36 is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the short arm of chromosome 1, characterized by borderline to mild intellectual disability, mild developmental delay, metopic craniosynostosis and mild craniofacial dysmorphism (incl. slopping forehead, bitemporal narrowing, blepharophimosis). Other associated abnormalities may include growth retardation, microcephaly, large hands, syndactyly, supernumerary ribs, rectal stenosis and/or anterior displacement of anus. Congenital heart malformations (e.g. atrial septal defect, patent ductus arteriosus) have also been reported.