Distal monosomy 15q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal monosomy 15q. ORPHA:1596

Distal monosomy 15q is a rare chromosomal anomaly syndrome characterized by pre- and postnatal growth restriction, developmental delay, variable degrees of intellectual disability, hand and foot anomalies (e.g. brachy-/clinodactyly, talipes equinovarus, nail hypoplasia, proximally placed digits) and mild craniofacial dysmorphism (incl. microcephaly, triangular face, broad nasal bridge, micrognathia). Neonatal lymphedema, heart malformations, aplasia cutis congenita, aortic root dilatation, and autistic spectrum disorder have also been reported.