Distal trisomy 18q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal trisomy 18q. ORPHA:1716

Distal trisomy 18q is a rare, partial autosomal trisomy characterized by a variable phenotype that includes hypotonia, motor delay, mild to severe intellectual disability, seizures, variable cerebral anomalies, finger/toe syndactyly, fifth finger clinodactyly, strabismus, short neck and dysmorphic facial features.