Monosomy 18q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Monosomy 18q. ORPHA:1600

Monosomy 18q is a partial deletion of the long arm of chromosome 18 characterized by highly variable phenotype, most commonly including hypotonia, developmental delay, short stature, growth hormone deficiency, hearing loss and external ear anomalies, intellectual disability, palatal defects, dysmorphic facial features, skeletal anomalies (foot deformities, tapering fingers, scoliosis) and mood disorders.