Distal 22q11.2 microdeletion syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal 22q11.2 microdeletion syndrome. ORPHA:261330

Distal 22q11.2 microdeletion syndrome is a rare chromosomal anomaly syndrome, resulting from the partial deletion of the long arm of chromosome 22, with a highly variable phenotype characterized by prematurity, pre- and post-natal growth retardation, developmental delay (particularly speech), mild intellectual disability, variable cardiac defects, and minor skeletal anomalies (such as clinodactyly). Dysmorphic features include prominent forehead, arched eyebrows, deep set eyes, narrow upslanting palpebral fissures, ear abnormalities, hypoplastic alae nasi, smooth philtrum, down-turned mouth, thin upper lip, retro/micrognatia and pointed chin. For certain very distal deletions, there is a risk of developing malignant rhabdoid tumours.