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Trisomy 4p

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Trisomy 4p. ORPHA:1738

Trisomy 4p is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the short arm of chromosome 4, with a highly variable phenotype, typically characterized by pre- and postnatal growth delay, psychomotor developmental delay and craniofacial dysmorphism (microcephaly, prominent glabelle, hypertelorism, enlarged ears with abnormal helix and antihelix, bulbous nose with flat or depressed nasal bridge, long philtrum, retrognathia with pointed chin). Additional features include skeletal (rocker bottom feet, arachnodactyly, camptodactyly) and renal malformations, cardiac defects, ocular abnormalities and abnormal genitalia in males.