Tetrasomy 9p

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Tetrasomy 9p. ORPHA:3310

Tetrasomy 9p is a rare autosomal anomaly characterized by pre- and postnatal growth retardation, psychomotor delay, mild to moderate intellectual disability, hypotonia, microcephaly, dysmorphic features (ocular hypertelorism, low-set, malformed ears, bulbous/beaked nose, microretrognathia, enophthalmos/micropthalmia, epicanthus, strabismus), cleft lip/palate, skeletal abnormalities (hypoplastic nails/distal phalanges, short stature, short neck, contractures), congenital heart defects, renal and urogenital malformations (renal hypoplasia, genital hypoplasia, cryptorchidism).