Monosomy 9p

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Monosomy 9p. ORPHA:261112

Monosomy 9p is a rare chromosomal anomaly characterized by psychomotor developmental delay, facial dysmorphism (trigonocephaly, midface hypoplasia, upslanting palpebral fissures, dysplastic small ears, flat nasal bridge with anteverted nostrils and long philtrum, micrognathia, choanal atresia, short neck), single umbilical artery, omphalocele, inguinal or umbilical hernia, genital abnormalities (hypospadia, cryptorchidism), muscular hypotonia and scoliosis.