Trisomy 12p

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Trisomy 12p. ORPHA:1699

A partial autosomal trisomy characterized by developmental delay and intellectual disability, generalized hypotonia, postnatal growth retardation, variable brain and heart anomalies and dysmorphic features, including frontal bossing, round face, full cheeks, low-set ears, broad nasal bridge, short nose with anteverted nares, long philtrum, thin upper lip vermilion, and everted, thick lower lip. Unspecific associated congenital anomalies have also been reported.