Distal trisomy 14q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal trisomy 14q. ORPHA:1705

Distal trisomy 14q is a rare, partial duplication of the long arm of chromosome 14 characterized by variable clinical features, most commonly including growth retardation and low birth weight, hypotonia, developmental delay, intellectual disability, short stature, microcephaly, facial dysmorphism (frontal bossing, hypertelorism, bulbous nose, micrognathia, sparse hair and eyebrows), congenital heart defects, spasticity and hyperreflexia.