Monosomy 13q14

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Monosomy 13q14. ORPHA:1587

Monosomy 13q14 is a rare chromosomal anomaly syndrome, resulting from a partial deletion of the long arm of chromosome 13, characterized by developmental delay, variable degrees of intellectual disability, retinoblastoma and craniofacial dysmorphism (incl. micro/dolichocephaly, high and broad forehead, prominent eyebrows, thick, anteverted ear lobes, short nose with a broad nasal bridge and bulbous tip, prominent philtrum, large mouth with thin upper lip and thick, everted lower lip). Other features reported include high birth weight, macrocephaly, pinealoma, hepatomegaly, inguinal hernia and cryptorchidism.