15q overgrowth syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 15q overgrowth syndrome. ORPHA:314585

15q overgrowth syndrome is a rare partial autosomal trisomy/tetrasomy characterized by facial dysmorphism (long thin face, prominent forehead, down-slanting palpebral fissures, prominent nose with broad nasal bridge, prominent chin), pre- and postnatal overgrowth, renal anomalies (e.g. horseshoe kidney, renal agenesis, hydronephrosis), mild to severe learning difficulties and behavioral abnormalities. Additional features may include craniosynostosis and macrocephaly.