17q12 microdeletion syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 17q12 microdeletion syndrome. ORPHA:261265

17q12 microdeletion syndrome is a rare chromosomal anomaly syndrome resulting from the partial deletion of the long arm of chromosome 17 characterized by renal cystic disease, maturity onset diabetes of the young type 5, and neurodevelopmental disorders, such as cognitive impairment, developmental delay (particularly of speech), autistic traits and autism spectrum disorder. Müllerian aplasia in females, macrocephaly, mild facial dysmorphism (high forehead, deep set eyes and chubby cheeks) and transient hypercalcaemia have also been reported.