Distal Xq28 microduplication syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal Xq28 microduplication syndrome. ORPHA:293939

Distal Xq28 microduplication syndrome is a rare, hereditary, syndromic intellectual disability characterized by cognitive impairment, behavioral and psychiatric problems, recurrent infections, atopic diseases, and distinctive facial features in males. Females are clinically asymptomatic or mildly affected, presenting mild learning difficulties and facial dysmorphism.