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Distal 17p13.1 microdeletion syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. [Distal 17p13.1 microdeletion syndrome](#). ORPHA:319171

Distal 17p13.1 microdeletion syndrome is a rare chromosomal anomaly syndrome characterized by mild global developmental delay/intellectual disability with poor to absent speech, dysmorphic features (long midface, retrognathia with overbite, protruding ears), microcephaly, failure to thrive, wide-based gait and a body posture with knee and elbow flexion and hands held in a midline.