Open Peer Review on Qeios

Tetrasomy 18p

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Tetrasomy 18p. ORPHA:3307

Tetrasomy 18p is a very rare structural chromosomal anomaly affecting multiple body systems and characterized clinically by craniofacial abnormalities, delayed development, cognitive impairment, changes in muscle tone, distinctive facial features, and rarely renal malformations.