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Distal monosomy 12p

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Distal monosomy 12p. ORPHA:280325*

Distal monosomy 12p is a rare partial autosomal monosomy characterized by language development delay with childhood apraxia of speech, mild intellectual disability, behavioural abnormalities (autistic spectrum disorder, attention deficit hyperactivity disorder, anxiety) and mildly dysmorphic nonspecific features. Additional clinical features may include muscular hypotonia and joint laxity, hernias and microcephaly.