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Intellectual disability-severe speech delay-mild dysmorphism syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Intellectual disability-severe speech delay-mild dysmorphism syndrome. ORPHA:391372*

Intellectual disability-severe speech delay-mild dysmorphism syndrome is a rare, genetic, syndromic intellectual disability disorder, with highly variable phenotype, typically characterized by mild to severe global development delay, severe speech and language impairment, mild to severe intellectual disability, dysphagia, hypotonia, relative to true macrocephaly, and behavioral problems that may include autistic features, hyperactivity, and mood lability. Facial gestalt typically features a broad, prominent forehead, hypertelorism, downslanting palpebral fissures, ptosis, a short bulbous nose with broad tip, thick vermilion border, wide, and open mouth with downturned corners. Brain, cardiac, urogenital and ocular malformations may be associated.