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X-linked intellectual disability-short stature-overweight syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. X-linked intellectual disability-short stature-overweight syndrome. ORPHA:457240*

X-linked intellectual disability-short stature-overweight syndrome is a multiple congenital anomalies syndrome characterized by borderline to severe intellectual disability, speech delay, short stature, elevated body mass index, a pattern of truncal obesity (reported in older males), and variable neurologic features (e.g. hypotonia, tremors, gait disturbances, behavioral problems, and seizure disorders). Less common manifestations include microcephaly, microorchidism and/or microphallus. Dysmorphic features have been reported in some patients but no consistent pattern has been noted.