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X-linked intellectual disability-acromegaly-hyperactivity syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. X-linked intellectual disability-acromegaly-hyperactivity syndrome. ORPHA:85327*

X-linked intellectual disability-acromegaly-hyperactivity syndrome is characterised by severe intellectual deficit, acromegaly and hyperactivity. The syndrome has been described in two half-brothers. Dysarthria, aggressive behaviour, a characteristic facies (an acromegalic and triangular face with a long nose) and macroorchidism were also present. The mother displayed moderate intellectual deficit and milder facial anomalies. Central nervous system anomalies were identified in the two boys: subarachnoid cysts and hyperdensity in the pontine region.