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Intellectual disability-short stature-hypertelorism syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Intellectual disability-short stature-hypertelorism syndrome. ORPHA:3074*

Intellectual disability-short stature-hypertelorism syndrome is a rare genetic syndromic intellectual disability characterized by short stature, mild to moderate intellectual disability, craniofacial dysmorphism (prominent broad 'square' forehead, hypertelorism, depressed nasal bridge, broad nasal tip and anteverted nares) and early hypotonia, typically present until infancy. There have been no further descriptions in the literature since 1991.