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Severe intellectual disability-short stature-behavioral abnormalities-facial dysmorphism syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Severe intellectual disability-short stature-behavioral abnormalities-facial dysmorphism syndrome. ORPHA:391307*

Severe intellectual disability-short stature-behavioral abnormalities-facial dysmorphism syndrome is a rare, genetic, syndromic intellectual disability disorder characterized by severe intellectual disability with limited or absent speech and language, short stature, acquired microcephaly, kyphoscoliosis or scoliosis, and behavioral disturbances that include hyperactivity, stereotypy and aggressiveness. Facial dysmorphism, that typically includes sloping forehead, mild synophrys, deep-set eyes, strabismus, anteverted large ears, prominent nose and dental malposition, is also characteristic.