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Intellectual disability-facial dysmorphism syndrome due to SETD5 haploinsufficiency

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Intellectual disability-facial dysmorphism syndrome due to SETD5 haploinsufficiency. ORPHA:404440*

Intellectual disability-facial dysmorphism syndrome due to SETD5 haploinsufficiency is a rare, syndromic intellectual disability characterized by intellectual disability of various severity, hypotonia, feeding difficulties, dysmorphic features, autism and behavioral issues. Growth retardation, congenital heart anomalies, gastrointestinal and genitourinary defects have been rarely associated.