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AHDC1-related intellectual disability-obstructive sleep apnea-mild dysmorphism syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. AHDC1-related intellectual disability-obstructive sleep apnea-mild dysmorphism syndrome. ORPHA:412069*

AHDC1-related intellectual disability-obstructive sleep apnea-mild dysmorphism syndrome is a rare, syndromic intellectual disability characterized by hypotonia, developmental delay, absent or severely delayed speech development, intellectual disability, obstructive sleep apnea, mild dysmorphic facial features and behavioral abnormalities. Epilepsy, ataxia and nystagmus have also been reported.