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Early-onset parkinsonism-intellectual disability syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Early-onset parkinsonism-intellectual disability syndrome. ORPHA:2379*

Early-onset parkinsonism with intellectual deficit is a basal ganglia disorder characterised by parkinsonian-type symptoms (postural changes, tremor, rigidity), megalencephaly and variable intellectual deficit. Other signs are frontal bossing, persistent frontal lobe reflexes, strabismus and seizures. It has been described in three generations of one family. Transmission is X-linked, and the gene is located on chromosomal region Xq27.3-qter.