

Open Peer Review on Qeios

Progressive myoclonic epilepsy type 5

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Progressive myoclonic epilepsy type 5. ORPHA:402082

A rare, genetic neurological disorder characterized by early-onset progressive ataxia associated with myoclonic seizures, generalized tonic-clonic seizures (which are often sleep-related), and normal to mild intellectual disability. Dysarthria, upward gaze palsy, sensory neuropathy, developmental delay and autistic disorder have also been associated.

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