

Open Peer Review on Qeios

Progressive myoclonic epilepsy type 8

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Progressive myoclonic epilepsy type 8. ORPHA:424027

A rare, genetic, neurological disorder characterized by childhood to adolescent-onset of action myoclonus, generalized tonic-clonic seizures, and slowly progressive, moderate to severe cognitive impairment that may lead to dementia. EEG reveals progressive slowing of background activity and epileptic abnormalities and brain MRI shows cerebellar and brainstem atrophy.

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