X-linked non progressive cerebellar ataxia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. X-linked non progressive cerebellar ataxia. ORPHA:314978

X-linked non progressive cerebellar ataxia is a rare hereditary ataxia characterized by delayed early motor development, severe neonatal hypotonia, non-progressive ataxia and slow eye movements, presenting normal cognitive abilities and absence of pyramidal signs. Frequently patients also manifest intention tremor, mild dysphagia, and dysarthria. Brain MRI reveals global cerebellar atrophy with absence of other malformations or degenerations of the central and peripheral nervous systems.