Autosomal recessive cerebellar ataxia-saccadic intrusion syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive cerebellar ataxia-saccadic intrusion syndrome. ORPHA:95434

Autosomal recessive cerebellar ataxia-saccadic intrusion syndrome is a rare hereditary ataxia characterized by a progressive cerebellar ataxia associated with disruption of visual fixation by saccadic intrusions (overshooting horizontal saccades with macrosaccadic oscillations and increased velocity of larger saccades). It presents with progressive gait, trunk and limb ataxia with pyramidal tract signs (increased tendon reflexes and Babinski sign), myoclonic jerks, fasciculations, cerebellar dysarthria, sensorimotor axonal neuropathy with impaired joint position, vibration, temperature, pain sensations, pes cavus, and saccadic intrusions with characteristic overshooting horizontal saccades, macrosaccadic oscillations, and increased velocity of larger saccades, without other eye movement disturbances.