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# Spinocerebellar ataxia type 43

INSERM

## Source

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

*Spinocerebellar ataxia type 43. ORPHA:497764*

Spinocerebellar ataxia type 43 is a rare autosomal dominant cerebellar ataxia type I disorder characterized by late adult-onset of slowly progressive cerebellar ataxia, typically presenting with balance and gait disturbances, in association with axonal peripheral neuropathy resulting in reduced/absent deep tendon reflexes and sensory impairment. Lower limb pain and amyotrophy may be present, as well as various cerebellar signs, including dysarthria, nystagmus, hypometric saccades and tremor.