Spinocerebellar ataxia with epilepsy

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Spinocerebellar ataxia with epilepsy. ORPHA:254881

Spinocerebellar ataxia with epilepsy is a rare, mitochondrial DNA maintenance syndrome characterized by cerebellar ataxia, sensory peripheral neuropathy, myoclonus, epilepsy, progressive cognitive impairment, late-onset ptosis and external ophthalmoplegia. Liver failure may also occur, most often in association with the use of antiepileptic drug sodium valproate.