Spinocerebellar Ataxia Type 12

National Cancer Institute

Source

National Cancer Institute. Spinocerebellar Ataxia Type 12. NCI Thesaurus. Code C154316.

An autosomal dominant sub-type of spinocerebellar ataxia caused by mutation(s) in the PPP2R2B gene, encoding serine/threonine-protein phosphatase 2A 55 kDa regulatory subunit B beta isoform. It presents with characteristic action tremors in the upper limbs, followed by other movement abnormalities.