# Spastic paraplegia-optic atrophy-neuropathy syndrome

INSERM

## Source

*INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Spastic paraplegia-optic atrophy-neuropathy syndrome. ORPHA:320406*

Spastic paraplegia-optic atrophy-neuropathy (SPOAN) syndrome is a rare, complex type of hereditary spastic paraplegia characterized by early-onset progressive spastic paraplegia presenting in infancy, associated with optic atrophy, fixation nystagmus, polyneuropathy occurring in late childhood/early adolescence leading to severe motor disability and progressive joint contractures and scoliosis. SPOAN syndrome is caused by mutations in the KLC2 gene (11q13.1), encoding kinesin light chain 2.