Spinal atrophy-ophthalmoplegia-pyramidal syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Spinal atrophy-ophthalmoplegia-pyramidal syndrome. ORPHA:1217

Spinal atrophy-ophthalmoplegia-pyramidal syndrome is a rare, bulbospinal muscular atrophy characterized by generalized neonatal hypotonia, progressive pontobulbar and spinal palsy, pyramidal signs, and deafness. External ophthalmoplegia and bilateral mydriasis are typical signs. There have been no further descriptions in the literature since 1994.