X-linked spinocerebellar ataxia type 3

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. X-linked spinocerebellar ataxia type 3. ORPHA:85297

X-linked spinocerebellar ataxia type 3 is a form of spinocerebellar degeneration characterized by onset in infancy of hypotonia, ataxia, sensorineural deafness, developmental delay, esotropia, and optic atrophy, and by a progressive course leading to death in childhood. It has been described one family with at least six affected males from five different sibships (connected through carrier females). It is transmitted as an X-linked recessive trait.