Spectrin-associated autosomal recessive cerebellar ataxia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Spectrin-associated autosomal recessive cerebellar ataxia. ORPHA:352403

Spectrin-associated autosomal recessive cerebellar ataxia is a rare, genetic neurological disease, due to SPTBN2 mutations, characterized by global development delay in infancy, followed by childhood-onset gait ataxia with limb dysmetria and dysdiadochokinesia, mild to severe intellectual disability, development of cerebellar atrophy, and abnormal eye movements (including a convergent squint, hypometric saccades, jerky pursuit movements and incomplete range of movement).