Autosomal recessive cerebellar ataxia-psychomotor delay syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive cerebellar ataxia-psychomotor delay syndrome. ORPHA:284271

A rare, hereditary, cerebellar ataxia disorder characterized by late-onset spinocerebellar ataxia, manifesting with slowly progressive gait disturbances, dysarthria, limb and truncal ataxia, and smooth-pursuit eye movement disturbance, associated with a history of psychomotor delay from childhood. Mild atrophy of the cerebellar vermis and hemispheres is observed on brain imaging.