Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to WWOX deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to WWOX deficiency. ORPHA:284282

Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to WWOX deficiency is a rare autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome characterized by early-childhood onset of cerebellar ataxia associated with generalized tonic-clonic epilepsy and psychomotor development delay, dysarthria, gaze-evoked nystagmus and learning disability. Other features in some patients include upper motor neuron signs with leg spasticity and extensor plantar responses, and mild cerebellar atrophy on brain MRI.