Autosomal recessive cerebellar ataxia-pyramidal signs-nystagmus-oculomotor apraxia syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive cerebellar ataxia-pyramidal signs-nystagmus-oculomotor apraxia syndrome. ORPHA:363429

Autosomal recessive cerebellar ataxia-pyramidal signs-nystagmus-oculomotor apraxia syndrome is a rare, genetic, slowly progressive neurodegenerative disease characterized by delayed psychomotor development beginning in infancy, mild to profound intellectual disability, gait and stance ataxia, pyramidal signs (hyperreflexia, extensor plantar responses), dysarthria, and ocular abnormalities (e.g. nystagmus, oculomotor apraxia, abduction deficits, esotropia, ptosis). Brain imaging reveals progressive, generalized cerebellar atrophy, mild ventriculomegaly and, in some, retrocerebellar cysts.