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Ataxia-intellectual disability-oculomotor apraxia-cerebellar cysts syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Ataxia-intellectual disability-oculomotor apraxia-cerebellar cysts syndrome. ORPHA:370022*

Ataxia-intellectual disability-oculomotor apraxia-cerebellar cysts syndrome is a rare neuro-ophthalmological disease characterized by nonprogressive cerebellar ataxia, delayed motor and language development and intellectual disability, in addition to ophthalmological abnormalities (e.g. oculomotor apraxia, strabismus, amblyopia, retinal dystrophy and myopia). Cerebellar cysts, cerebellar dysplasia and cerebellar vermis hypoplasia, seen on magnetic resonance imaging, are also characteristic of the disease.