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# Ataxia-hypogonadism-choroidal dystrophy syndrome

INSERM

## Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Ataxia-hypogonadism-choroidal dystrophy syndrome. ORPHA:1180*

Ataxia-hypogonadism-choroidal dystrophy syndrome is a very rare autosomal recessive, slowly progressive neurodegenerative disorder characterized by the triad of cerebellar ataxia (that generally manifests at adolescence or early adulthood), chorioretinal dystrophy, which may have a later onset (up to the fifth-sixth decade) leading to variable degrees of visual impairment, and hypogonadotropic hypogonadism (delayed puberty and lack of secondary sex characteristics). Ataxia-hypogonadism-choroidal dystrophy syndrome belongs to a clinical continuum of neurodegenerative disorders along with the clinically overlapping cerebellar ataxia-hypogonadism syndrome (see this term).