DISEASE:
Ataxia-hypogonadism-choroidal dystrophy syndrome

NAME:	Ataxia-hypogonadism-choroidal dystrophy syndrome
DESCRIPTION:	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).
ORPHACODE:	1180
SYNONYMS:	Boucher-Neuhäuser syndrome
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	PNPLA6
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