

DISEASE:
Cerebellar ataxia-hypogonadism syndrome

NAME:	Cerebellar ataxia-hypogonadism syndrome
DESCRIPTION:	Cerebellar ataxia-hypogonadism syndrome is a very rare autosomal recessive neurodegenerative disorder characterized by the combination of progressive cerebellar ataxia with onset from early childhood to the fourth decade, and hypogonadotropic hypogonadism (delayed puberty and lack of secondary sex characteristics). Cerebellar ataxia-hypogonadism syndrome belongs to a clinical continuum of neurodegenerative disorders along with clinically overlapping disorders such as ataxia-hypogonadism-choroidal dystrophy syndrome (see this term).
ORPHACODE:	1173
SYNONYMS:	Gordon-Holmes syndrome Luteinizing hormone-releasing hormone deficiency with ataxia
XREF(S):	Orphanet OMIM OMIM ICD-10
ANALYTE(S):	PNPLA6 RNF216
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