Opis choroby *

Definicja

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).

Dane

Klasyfikacja Synonimy

Choroba Gordon-Holmes syndrome

Niedobór gonadoliberyny z ataksją

Zespół Gordona Holmesa

Luteinizing hormone-releasing hormone

deficiency with ataxia

Kod ORPHA

1173

Kod OMIM

Kod ICD10

605672 G11.8

Kod ICD11

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*Źródło

orphanet