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
Choroba

Synonimy
Gordon-Holmes syndrome
Niedobór gonadoliberyny z ataksją
Zespół Gordona Holmesa
Luteinizing hormone-releasing hormone deficiency with ataxia

Kod ORPHA
1173

Kod OMIM
605672

Kod ICD10
G11.8

Kod ICD11
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*Źródło

orphanet