

Opis choroby *

Definicja

Intellectual disability-myopathy-short stature-endocrine defect syndrome is a rare congenital myopathy syndrome characterized by nonprogressive myopathy (manifesting with mild facial and generalized weakness, bilateral ptosis, and severe lumbar lordosis), severe intellectual disability, short stature, and sexual infantilism (due to hypogonadotropic hypogonadism). The presence of a small pituitary fossa was also noted. There have been no further descriptions in the literature since 1985.

Dane

Klasyfikacja

Choroba

Synonimy

Chudley-Rozdilsky syndrome

Zespół Chudley'a i Rozdilsky'ego

Kod ORPHA

3068

Kod OMIM

253320

Kod ICD10

Q87.8

Kod ICD11

LD90

*Źródło

orphanet