

## 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