

## Opis choroby \*

### Definicja

A rare, genetic, syndromic intellectual disability disorder characterized by non-progressive, congenital, marked, central hypotonia, severe psychomotor delay and intellectual disability, chronic constipation, distended abdomen, abnormal dermatoglyphics, delayed and dysharmonic skeletal maturation, and preponderance of type 2 larger-sized muscle fibers. Additional features include narrow and high-arched palate, prominent nasal root, long philtrum, and open mouth with drooling, as well as variably present cryptorchidism, hypertelorism, and tapered fingers. Seizures and/or an abnormal electroencephalograph may also be associated. There have been no further descriptions in the literature since 1994.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Dysharmonic skeletal maturation-muscular fiber disproportion syndrome  
Nieharmonijne dojrzewanie szkieletu - dysproporcja włókien mięśniowych

#### Kod ORPHA

3010

#### Kod OMIM

600096

#### Kod ICD10

Q87.8

#### Kod ICD11

-

---

#### [\\*Źródło](#)

orphanet