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 assoicated. 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
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*Źródło

orphanet