

Zespół Qazi i Markouizosa

Kod Orpha: 3010 Kod OMIM: 600096

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

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[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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