Opis choroby *

Definicja

Richieri Costa-da Silva syndrome is a rare, genetic, myotonic syndrome characterized by childhood onset of progressive and severe myotonia (with generalized muscular hypertrophy and progressive impairment of gait), short stature, skeletal abnormalities (including pectus carinatum, short, wedge-shaped thoracolumbar vertebrae, kyphoscoliosis, genu valgum, irregular femoral epiphyses), and mild to moderate intellectual deficiency. No facial dysmorphism nor joint limitation is associated. There have been no further descriptions in the literature since 1984.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Myotonia-intellectual disability-skeletal anomalies syndrome
Miotonia - niepełnosprawność intelektualna - wady szkieletu

Kod ORPHA
3101

Kod OMIM
255710

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet