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 Synonimy

Zespół wad wrodzonych Myotonia-intellectual disability-skeletal

anomalies syndrome

Miotonia - niepełnosprawność intelektualna -

wady szkieletu

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 3101
 255710
 Q87.8

Kod ICD11

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<u>*Źródło</u>

orphanet