Opis choroby *

Definicja

Congenital muscular dystrophy without intellectual disability is a rare, genetic, congenital muscular dystrophy due to dystroglycanopathy disorder characterized by a wide phenotypic spectrum which includes hypotonia and muscular weakness present at birth or early infancy, delayed or arrested motor development, and normal intellectual abilities with normal (or only mild abnormalities) neuroimaging studies. Feeding difficulties, joint and spinal deformities, and respiratory insufficiency may be associated. Decreased alpha-dystroglycan on immunohistochemical muscle staining and elevated serum creatine kinase are observed.

Dane

Klasyfikacja
Choroba

Synonimy
CMD without intellectual disability
CMD bez niepełnosprawności intelektualnej
CMD-no MR
CMD-no MR
Congenital muscular dystrophy-dystroglycanopathy without intellectual disability

Kod ORPHA
370980

Kod OMIM
613152

Kod ICD10
G71.2

Kod ICD11
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*Źródło

orphanet