

Opis choroby *

Definicja

Creatine deficiency syndrome (CDS) comprises a group of inborn errors of creatine metabolism, characterized by a global developmental delay, intellectual disability and associated neurological (seizures, movement disorders, myopathy) and behavioral manifestations. CDS includes two creatine biosynthesis disorders; guanidinoacetate methyltransferase deficiency and L- Arginine: glycine amidinotransferase deficiency, as well as X-linked creatine transporter deficiency.

Dane

Klasyfikacja	Synonimy
Grupa fenomenów	CCDS CDS Zespół niedoboru kreatyny mózgowej CDS Cerebral creatine deficiency syndrome

Kod ORPHA	Kod OMIM	Kod ICD10
79172	-	E72.8

Kod ICD11

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*Źródło

orphanet