

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

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

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|------------------|-----------------|------------------|
| Kod ORPHA | Kod OMIM | Kod ICD10 |
| 79172 | - | E72.8 |

Kod ICD11

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*Źródło

orphanet