

Opis choroby *

Definicja

Recurrent metabolic encephalomyopathic crises-rhabdomyolysis-cardiac arrhythmia-intellectual disability syndrome is a rare, genetic, neurodegenerative disease characterized by episodic metabolic encephalomyopathic crises (of variable frequency and severity which are frequently precipitated by an acute illness) which manifest with profound muscle weakness, ataxia, seizures, cardiac arrhythmias, rhabdomyolysis with myoglobinuria, elevated plasma creatine kinase, hypoglycemia, lactic acidosis, increased acylcarnitines and a disorientated or comatose state. Global developmental delay, intellectual disability and cortical, pyramidal and cerebellar signs develop with subsequent progressive neurodegeneration causing loss of expressive language and varying degrees of cerebral atrophy.

Dane

Klasyfikacja

Choroba

Synonimy

TANGO2-related metabolic encephalopathy-
arrhythmia syndrome
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arrhythmia syndrome

Kod ORPHA

480864

Kod OMIM

616878

Kod ICD10

G31.8

Kod ICD11

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[*Źródło](#)

orphanet