

Opis choroby *

Definicja

A rare genetic disease characterized by the association of epilepsy, ataxia, sensorineural hearing impairment, and renal tubulopathy. Patients present in infancy with generalized seizures, cerebellar dysfunction (including gait ataxia, intention tremor, and dysdiadochokinesis), and variable developmental delay and sensorineural hearing loss. Laboratory studies show persistent hypokalemic metabolic acidosis with hypomagnesemia. Additional reported neurologic features include brisk deep tendon reflexes, ankle clonus, extensor plantar responses, or nystagmus.

Dane

Klasyfikacja

Choroba

Synonimy

Epilepsy-ataxia-sensorineural deafness-tubulopathy syndrome

Drgawki - głuchota czuciowo-nerwowa - ataksja - niepełnosprawność intelektualna - brak równowagi elektrolitowej

Zespół SeSAME

Epilepsy-ataxia-sensorineural hearing loss-tubulopathy syndrome

SeSAME syndrome

Seizures-sensorineural deafness-ataxia-intellectual disability-electrolyte imbalance syndrome

Seizures-sensorineural hearing loss-ataxia-intellectual disability-electrolyte imbalance syndrome

Kod ORPHA

199343

Kod OMIM

612780

Kod ICD10

G40.4

Kod ICD11

LD2H.Y

[*Źródło](#)

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