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

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*Źródło

orphanet