Opis choroby *

Definicja

A rare genetic disease characterized by juvenile-onset insulin-dependent diabetes mellitus associated with central and peripheral nervous system abnormalities with variable onset between infancy and adolescence. Neurological manifestations include combined cerebellar and afferent ataxia, sensorineural hearing loss, pyramidal tract signs, and demyelinating sensorimotor peripheral neuropathy. Hypothyroidism has been reported in some patients. Brain imaging may show generalized cerebral atrophy.

Dane

Klasyfikacja
Choroba

Synonimy
Combined cerebellar and peripheral ataxia-deafness-diabetes mellitus syndrome
Zespół złożonej ataksji móżdżkowej i obwodowej, utraty słuchu i cukrzycy
Combined cerebellar and peripheral ataxia-hearing loss-diabetes mellitus syndrome

Kod ORPHA
445062

Kod OMIM
616192

Kod ICD10
G31.8

Kod ICD11
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*Źródło

orphanet