

Zespół poliendokrynopatii i polineuropatii

Kod Orpha: 453533 Kod OMIM: 616113

Opis choroby *

Definicja

A rare genetic disease characterized by childhood onset of multiple endocrine manifestations in combination with central and peripheral nervous system abnormalities. Reported signs and symptoms include postnatal growth retardation, moderate intellectual disability, hypogonadotropic hypogonadism, insulin-dependent diabetes mellitus, central hypothyroidism, demyelinating sensorimotor polyneuropathy, and cerebellar and pyramidal signs. Progressive hearing loss and a hypoplastic pituitary gland have also been described. Brain imaging shows moderate white matter abnormalities.

Dane

Klasyfikacja

Choroba

Kod ORPHA

453533

Kod OMIM

616113

Kod ICD10

E34.8

Kod ICD11

-

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.