Opis choroby *

Definicja

A rare genetic leukodystrophy identified in families of Ashkenazi Jewish descent, characterized by infancy onset of severe global developmental delay with very limited or absent speech and sometimes complete absence of motor development, hypotonia, spasticity, and acquired microcephaly. Seizures, hearing loss, visual impairment, and autonomic dysfunction have also been described. Brain imaging shows delayed myelination and other white matter abnormalities.

Dane

Klasyfikacja
Choroba

Synonimy
VPS11-related autosomal recessive hypomyelinating leukoencephalopathy
Autosomalna recesywna leukoencefalopatia hipomielinizacyjna zależna od VPS11

Kod ORPHA
466934

Kod OMIM
616683

Kod ICD10
G93.8

Kod ICD11
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*Źródło

orphanet