Opis choroby *

Definicja

A rare neurometabolic disorder due to serine deficiency characterized by neonatal to infantile onset of global developmental delay, postnatal microcephaly and intellectual disability, which may be associated with slowly progressive spastic tetraplegia mainly affecting the lower extremities, seizures, and brain MRI findings including thin corpus callosum, delayed myelination and cerebral atrophy. Additional symptoms include brisk deep tendon reflexes, extensor plantar responses, behavioral abnormalities (such as irritability, hyperactivity, sleep disorder), abnormal hand movements and stereotypy.

Dane

Klasyfikacja
Choroba

Synonimy
ASCT1 deficiency
Niedobór ASCT1
Zespół kwadriplegii spastycznej, cienkego ciała modzelowatego i postępującej mikrocefalii postnatalnej
Spastic quadriplegia-thin corpus callosum-progressive postnatal microcephaly syndrome

Kod ORPHA
447997

Kod OMIM
616657

Kod ICD10
Q02

Kod ICD11
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*Źródło

orphanet