

## 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, cienkiego 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