## Opis choroby *

Definicja

A rare disorder of manganese transport characterized by progressive movement disorder and elevated blood manganese levels. Patients present in infancy or early childhood with loss of motor milestones, rapidly progressive dystonia, spasticity, bulbar dysfunction, and parkinsonism, resulting in loss of independent ambulation. Cognition may be impaired but is generally better preserved than motor function. Additional manifestations include abnormal head growth and skull deformities. Brain MRI shows abnormalities of the basal ganglia, variably also of other brain regions.

Dane

**Klasyfikacja**
Choroba

**Kod ORPHA**
521406

**Kod OMIM**
617013

**Kod ICD10**
E83.8

**Kod ICD11**
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*Źródło

orphanet