

## Opis choroby \*

### Definicja

A rare disorder of manganese transport characterized by childhood onset of extrapyramidal movement disorder (including dystonia, tremor, and bradykinesia), liver cirrhosis, polycythemia, and hypermanganesemia. Cases with spastic paraparesis without extrapyramidal dysfunction have also been reported. Cognitive functions are preserved. Brain imaging findings are consistent with deposition of manganese in the basal ganglia, dentate nucleus, brain stem, and anterior pituitary.

### Dane

### Klasyfikacja

Choroba

#### Kod ORPHA

309854

#### Kod OMIM

613280

#### Kod ICD10

E88.8

#### Kod ICD11

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### \*Źródło

orphanet