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