Opis choroby *

Definicja

Microcephaly-complex motor and sensory axonal neuropathy syndrome is an extremely rare subtype of hereditary motor and sensory neuropathy characterized by severe, rapidly-progressing, distal, symmetric polyneuropathy and microcephaly (which can be evident in utero) with intact cognition. Clinically it presents with delayed motor development, hypotonia, absent or reduced deep tendon reflexes, progressive muscle wasting and weakness and scoliosis.

Dane

Klasyfikacja
Choroba

Kod ORPHA
423894

Kod OMIM
-

Kod ICD10
G60.0

Kod ICD11
-

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*Źródło

orphanet