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