Opis choroby *

Definicja

A rare, genetic, autosomal recessive axonal hereditary motor and sensory neuropathy disease characterized by prenatal onset of a severe sensorimotor axonal polyneuropathy (reflected by reduced fetal movement and polyhydramnios), manifesting, at birth, with respiratory failure requiring mechanical ventilation, profound muscular hypotonia, rapidly progressing distal muscle weakness, and absent deep tendon reflexes, in the absence of contractures, leading to death before 8 months of age. Neuropathological findings show severe loss of large- and medium-sized myelinated fibers without signs of demyelination.

Dane

Klasyfikacja
Choroba

Kod ORPHA
538096

Kod OMIM
604431

Kod ICD10
G60.0

Kod ICD11
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*Źródło

orphanet