

Opis choroby *

Definicja

Lethal congenital contracture syndrome type 1 is a rare, genetic arthrogryposis syndrome characterized by total fetal akinesia (detectable since the 13th week of gestation) accompanied by hydrops, micrognathia, pulmonary hypoplasia, pterygia and multiple joint contractures (usually flexion contractures in the elbows and extension in the knees), leading invariably to death before the 32nd week of gestation. Lack of anterior horn motoneurons, severe atrophy of the ventral spinal cord and severe skeletal muscle hypoplasia are characteristic neuropathological findings, with no evidence of other organ structural anomalies.

Dane

Klasyfikacja

Zespół wad wrodzonych Herva disease

Synonimy

Choroba Herva

LCCS1

Zespół mnogich przykurczów, typu fińskiego

LCCS1

Multiple contracture syndrome, Finnish type

Kod ORPHA

1486

Kod OMIM

253310

Kod ICD10

Q68.8

Kod ICD11

LD26.4Y

[*Źródło](#)

orphanet