

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	Synonimy
Zespół wad wrodzonych	Herva disease
	Choroba Herva
	LCCS1
	Zespół mnogich przykurczów, typu fińskiego
	LCCS1
	Multiple contracture syndrome, Finnish type

Kod ORPHA	Kod OMIM	Kod ICD10
1486	253310	Q68.8

Kod ICD11
LD26.4Y

*Źródło

orphanet