

Opis choroby *

Definicja

Lethal congenital contracture syndrome type 2 is a rare arthrogyposis syndrome characterized by multiple congenital contractures (typically extended elbows and flexed knees), micrognathia, anterior horn cell degeneration, skeletal muscle atrophy (mainly in the lower limbs), presence of a markedly distended urinary bladder and absence of hydrops, pterygia and bone fractures. Other craniofacial (e.g. cleft palate, facial palsy) and ocular (e.g. anisocoria, retinal detachment) anomalies may be additionally observed. The disease is usually neonatally lethal however, survival into adolescence has been reported.

Dane

Klasyfikacja

Zespół wad wrodzonych LCCS2

Synonimy

LCCS2

Zespół mnogich przykurczy stawowych, typ izraelsko-beduiński

Multiple contracture syndrome, Israeli-Bedouin type

Kod ORPHA

137776

Kod OMIM

607598

Kod ICD10

Q68.8

Kod ICD11

LD26.4Y

*Źródło

orphanet