

Opis choroby *

Definicja

Fetal akinesia-cerebral and retinal hemorrhage syndrome is a rare, lethal, congenital myopathy syndrome characterized by decreased fetal movements and polyhydramnios in utero and the presence of akinesia, severe hypotonia with respiratory insufficiency, absent reflexes, joint contractures, skeletal abnormalities with thin ribs and bones, intracranial and retinal hemorrhages and decreased birth weight in the neonate.

Dane

Klasyfikacja

Choroba

Synonimy

LCCS5

Lethal congenital contracture syndrome type 5

LCCS5

Lethal congenital contracture syndrome type 5

Kod ORPHA

363409

Kod OMIM

615368

Kod ICD10

G71.2

Kod ICD11

-

*Źródło

orphanet