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Fetal akinesia-cerebral and retinal hemorrhage syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Fetal akinesia-cerebral and retinal hemorrhage syndrome. ORPHA:363409*

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.