DISEASE:
Fetal akinesia-cerebral and retinal hemorrhage syndrome

NAME:	Fetal akinesia-cerebral and retinal hemorrhage syndrome
DESCRIPTION:	Fetal akinesia-cerebral and retinal hemorrhage syndrome is a rare, lethal, congenital myopathy syndrome characterized by decreased fetal movements and polyhydraminos 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.
ORPHACODE:	363409
SYNONYMS:	LCCS5 Lethal congenital contracture syndrome type 5
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	DNM2
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