Opis choroby *

Definicja

Lethal fetal cerebrorenogenitourinary agenesis/hypoplasia syndrome is a rare, genetic developmental defect during embryogenesis malformation syndrome characterized by intrauterine growth restriction, flexion arthrogryposis of all joints, severe microcephaly, renal cystic dysplasia/agenesis/hypoplasia and complex malformations of the brain (cerebral and cerebellar hypoplasia, vermis, corpus callosum and/or occipital lobe agenesis, with or without arhinencephaly), as well as of the genitourinary tract (ureteral agenesis/hypoplasia, uterine hypoplasia and/or vaginal atresia), leading to fetal demise.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
439897

Kod OMIM
616258

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet