Opis choroby *

Definicja

Kagami-Ogata syndrome is a rare genetic disease characterized by polyhydramnios (mostly due to placentomegaly), fetal macrosomia, abdominal wall defects, skeletal abnormalities (including bell-shaped thorax, coat-hanger appearance of the ribs and decreased mid to wide thorax diameter ratio in infancy), feeding difficulties and impaired swallowing, dysmorphic features (hairy forehead, full cheeks, protruding philtrum, micrognathia), developmental delay and intellectual disability. Additional features may include kyphoskoliosis, joint contractures, diastasis recti, muscular hypotonia. There is increased risk of hepatoblastoma.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
KOS
MCA z powodu ekspresji matczynego wadliwego genu 14q32.2

Kod ORPHA
254519

Kod OMIM
608149

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet