Opis choroby *

Definicja

A rare genetic lethal multiple congenital anomalies/dysmorphic syndrome characterized by mid-gestation lethality and features of a ciliopathy. Clinical manifestations include hydrocephalus, cerebellar vermis hypoplasia, corpus callosum agenesis, duodenal atresia, gastrointestinal malrotation, bilateral renal hypoplasia, and dysmorphic craniofacial features (such as microcephaly, hypertelorism, low-set ears, prominent nose, short columella, cleft palate, micrognathia, and wide mouth).

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
444069

Kod OMIM
243605

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet