Opis choroby *

Definicja

A rare genetic lethal multiple congenital anomalies/dysmorphic syndrome characterized by early intrauterine growth retardation, generalized edema, craniofacial dysmorphism (such as microcephaly, brachycephaly, frontal bossing, hypertelorism, short palpebral fissures, or absent nasal bone), cerebellar hypoplasia, sex reversal in male fetuses, congenital heart defects (including septal and valve defects and cardiomegaly), and late fetal loss.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
580933

Kod OMIM
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Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet