Opis choroby *

Definicja

A rare, multiple congenital anomalies/dysmorphic syndrome characterized by microcephaly, intellectual disability, seizures, and congenital heart defects (e.g. atrial/ventricular septal defect, hypoplastic aortic arch with persistent ductus arteriosus). Additional manifestations include mild hypothyroidism, skeletal abnormalities, micropenis, delayed psychomotor development, dysmorphic facial features (including epicanthus, depressed nasal bridge, prominent antitragus), and pulmonary vascular occlusive disease. There have been no further descriptions in the literature since 1989.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
2519

Kod OMIM
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Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet