

## 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