

## Opis choroby \*

### Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by global developmental delay, intellectual disability, short stature, skeletal abnormalities (such as brachydactyly and vertebral anomalies), obesity, cardiac, respiratory, and genitourinary anomalies, and dysmorphic facial features (including coarse facies, thick eyebrows, synophrys, hypertelorism, short, upturned nose, and long philtrum). Additional reported manifestations are microcephaly, hearing impairment, cataract, and gastroesophageal reflux.

### Dane

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych CHOPS syndrome	Zespół CHOPS

<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
444077	616368	Q87.8

**Kod ICD11**  
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### \*Źródło

orphanet