

## **Opis choroby \***

### Definicja

A partial autosomal trisomy characterized by developmental delay, intellectual disability, prenatal and postnatal growth retardation, congenital heart, genitourinary and skeletal anomalies, and dysmorphic facial features, including high and broad forehead, hypertelorism, upslanting palpebral fissures, broad nose, dysplastic and low set ears, micrognathia. Phenotypic features vary in relation to the duplication size.

### Dane

#### **Klasyfikacja**

Zespół wad wrodzonych Duplication 8q  
Duplikacja 8q

#### **Kod ORPHA**

1752

#### **Kod OMIM**

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#### **Kod ICD10**

Q92.2

#### **Kod ICD11**

LD41.70

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#### \*Źródło

orphanet