

## **Opis choroby \***

### Definicja

A very rare, chromosomal anomaly characterized by an intrauterine and postnatal growth retardation, short stature, developmental delay, learning difficulties, hearing loss, hypermetropia, and a recognisable facial dysmorphism including prominent forehead, long, myopathic facies, fine eyebrows, small mouth and micrognathia.

### Dane

#### **Klasyfikacja**

Zespół wad wrodzonych Del(1)(p35.2)

Del(1)(p35.2)

Delekcja 1p35.2

Monosomia 1p35.2

Deletion 1p35.2

Monosomy 1p35.2

#### **Synonimy**

#### **Kod ORPHA**

456298

#### **Kod OMIM**

-

#### **Kod ICD10**

Q93.5

#### **Kod ICD11**

-

---

#### \*Źródło

orphanet