

## Opis choroby \*

### Definicja

A rare, genetic, syndromic intellectual disability disorder characterized by mild to moderate intellectual disability, facial dysmorphism (including a long face, deep-set eyes, narrow-based, broad nose with nostril colobomata, mandibular prognathism), hypergonadotrophic hypogonadism, eunuchoid habitus, diabetes mellitus type 1, and epilepsy. There have been no further descriptions in the literature since 1990.

### Dane

### Klasyfikacja

Zespół wad wrodzonych

#### Kod ORPHA

3044

#### Kod OMIM

249599

#### Kod ICD10

Q87.8

#### Kod ICD11

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### \*Źródło

orphanet