

## Opis choroby \*

### Definicja

A rare genetic syndromic intellectual disability characterized by moderate to severe intellectual deficiency, language deficit (completely absent or significantly impaired speech), and distinctive facial dysmorphism (long face, straight eyebrows, and, less frequently, low-set ears and café-au-lait spots). Additional, variably observed features include motor delays, behavioral difficulties, and seizures.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Intellectual disability-loss of expressive language-facial dysmorphism syndrome  
Zespół niepełnosprawności intelektualnej, utraty języka ekspresywnego i dysmorfii twarzy

#### Kod ORPHA

436151

#### Kod OMIM

616083

#### Kod ICD10

Q87.0

#### Kod ICD11

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#### \*Źródło

orphanet