

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