

Opis choroby *

Definicja

A rare, genetic, syndromic intellectual disability characterized by developmental delay, mild to moderate intellectual disability, low birth weight, moderate to severe short stature, microcephaly and variable hypergonadotropic hypogonadism. Mild facial dysmorphism include upslanted palpebral fissures and prominent nasal bridge.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

163976

Kod OMIM

301030

Kod ICD10

Q87.8

Kod ICD11

LD90

*Źródło

orphanet