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 dismorfism 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