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