

Opis choroby *

Definicja

X-linked intellectual disability, Seemanova type is characterised by microcephaly, intellectual deficit, growth retardation and hypogonadism. It has been described in four boys from one family. A characteristic facies and ophthalmologic anomalies were also present and included microphthalmia, microcornea and cataract. Transmission is X-linked.

Dane

Klasyfikacja

Choroba

Kod ORPHA

85323

Kod OMIM

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Kod ICD10

Q87.8

Kod ICD11

LD20.2

*Źródło

orphanet