

Opis choroby *

Definicja

Microcephaly-microcornea syndrome, Seemanova type is characterised by microcephaly and brachycephaly, eye anomalies (microphthalmia, microcornea, congenital cataract), hypogenitalism, severe intellectual deficit, growth retardation and progressive spasticity. It has been described in two patients (a male and his sister's son). Both patients also presented with facial dysmorphism, including upslanting palpebral fissures, epicanthal folds, highly arched palate, microstomia, and retrognathia. This syndrome is transmitted as an X-linked trait.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych Seemanova-Lesny syndrome	Zespół Seemanova i Lesny

Kod ORPHA	Kod OMIM	Kod ICD10
2528	-	Q87.8

Kod ICD11
LD20.2

*Źródło

orphanet