Microcephaly-microcornea syndrome, Seemanova type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Microcephaly-microcornea syndrome, Seemanova type. ORPHA:2528

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.