

Open Peer Review on Qeios

X-linked intellectual disability, Seemanova type

INSFRM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. X-linked intellectual disability, Seemanova type. ORPHA:85323

X-linked intellectual disability, Seemanova type is characterised by microcephaly, intellectual deficit, growth retardation and hypogenitalism. 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.

Qeios ID: CRK1IU · https://doi.org/10.32388/CRK1IU