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X-linked intellectual disability, Schimke type

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. X-linked intellectual disability, Schimke type. ORPHA:85285*

X-linked mental retardation, Schimke type, is characterised by intellectual deficit, growth retardation with short stature, deafness and ophthalmoplegia. Choreoathetosis with muscle spasticity generally appears during childhood. It has been described in four boys, three of whom were from the same family. Transmission is X-linked.