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X-linked intellectual disability, Shashi type

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. X-linked intellectual disability, Shashi type. ORPHA:85286*

X-linked intellectual disability, Shashi type is characterised by moderate intellectual deficit, obesity, macroorchidism and a characteristic facies (large ears, a prominent lower lip and puffy eyelids). It has been described in nine boys from two families. Transmission is X-linked and the causative gene has been localised to the q21.3-q27 region of the X chromosome.