Syndromic X-linked intellectual disability due to JARID1C mutation

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Syndromic X-linked intellectual disability due to JARID1C mutation. ORPHA:85279

Syndromic X-linked intellectual disability due to JARID1C mutation is characterised by mild to severe intellectual deficit associated with variable clinical manifestations including spasticity, cryptorchidism, maxillary hypoplasia, alopecia areata, epilepsy, short stature, impaired speech and behavioural problems. To date, it has been described in less than 15 families. Transmission is X-linked recessive and the syndrome is caused by mutations in the JARID1C (SMCX) gene encoding a JmjC-domain protein with histone demethylase activity.