X-linked intellectual disability, Stevenson type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. X-linked intellectual disability, Stevenson type. ORPHA:85325

X-linked intellectual disability, Stevenson type is characterised by intellectual deficit, hypotonia, absent deep tendon reflexes, tapered fingers and excessive fingerprint arches, genu valgum, a characteristic face and small teeth. It has been described in four males from two generations of one family. The causative gene appears to be located in the q13 region of the X chromosome.