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X-linked intellectual disability-hypotonia-facial dysmorphism-aggressive behavior syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. X-linked intellectual disability-hypotonia-facial dysmorphism-aggressive behavior syndrome. ORPHA:85329*

X-linked intellectual disability-hypotonia-facial dysmorphism-aggressive behavior syndrome is characterised by severe intellectual deficit, hypotonia, mild facial dysmorphism, and aggressive behaviour. It has been described in 10 male members spanning four generations of one family. The facial dysmorphism includes a high forehead, prominent ears, and a small pointed chin. Height and head circumference are reduced. This disorder is transmitted as an X-linked recessive trait and the causative gene maps to Xp22.