Atkin-Flaitz syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Atkin-Flaitz syndrome. ORPHA:1193

Atkin-Flaitz syndrome is characterised by moderate to severe intellectual deficit, short stature, macrocephaly, and characteristic facies. It has been described in 11 males and three females from three successive generations of the same family. The males also presented with postpubertal macroorchidism. Transmission is X-linked.