# X-linked spasticity-intellectual disability-epilepsy syndrome

INSERM

## Source

*INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. X-linked spasticity-intellectual disability-epilepsy syndrome. ORPHA:3175*

This syndrome is characterised by myoclonic epilepsy with generalised spasticity and intellectual deficit. It has been described in six males from two generations of one family. Transmission appears to be X-linked recessive and the syndrome is caused by mutations in the aristaless-related homeobox gene (ARX, Xp22.13).