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X-linked intellectual disability-epilepsy-progressive joint contractures-dysmorphism syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. X-linked intellectual disability-epilepsy-progressive joint contractures-dysmorphism syndrome. ORPHA:85319*

X-linked intellectual disability-epilepsy-progressive joint contractures-dysmorphism syndrome is characterised by intellectual deficit, epilepsy, facial dysmorphism and progressive joint contractures. It has been described in two boys. Hypotonia and feeding problems at birth were also reported. The mode of transmission is X-linked.