Open Peer Review on Qeios

Spinocerebellar ataxia-dysmorphism syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Spinocerebellar ataxia-dysmorphism syndrome. ORPHA:1185

Spinocerebellar ataxia-dysmorphism is marked by characteristic facies associated with dysarthria, delayed psychomotor development, ataxia, scoliosis and foot deformities. Three cases have been described and transmission appears to be autosomal recessive.