Open Peer Review on Qeios

Polyneuropathy-intellectual disabilityacromicria-premature menopause syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Polyneuropathy-intellectual disability-acromicria-premature menopause syndrome. ORPHA:2928

Polyneuropathy-intellectual disability-acromicria-premature menopause syndrome is a rare genetic syndromic intellectual disability characterized by intellectual disability, polyneuropathy, short stature and short limbs, brachydactyly, and premature ovarian insufficiency. Only one familial case with three affected females was described and there have been no further descriptions in the literature since 1971.