Open Peer Review on Qeios

Autosomal recessive spastic paraplegia type 45

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive spastic paraplegia type 45. ORPHA:320396

Autosomal recessive spastic paraplegia type 45 is a rare, pure or complex form of hereditary spastic paraplegia characterized by onset in infancy of progressive lower limb spasticity, abnormal gait, increased deep tendon reflexes and extensor plantar responses, that may be associated with intellectual disability. Additional signs, such as contractures in the lower limbs, amyotrophy, clubfoot and optic atrophy, have also been reported.