Autosomal recessive spastic paraplegia type 54

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive spastic paraplegia type 54. ORPHA:320380

Autosomal recessive spastic paraplegia type 54 (SPG54) is a rare, complex form of hereditary spastic paraplegia characterized by the onset in early childhood of progressive spastic paraplegia associated with cerebellar signs, short stature, delayed psychomotor development, intellectual disability and, less commonly, foot contractures, dysarthria, dysphagia, strabismus and optic hypoplasia. SPG54 is caused by mutations in the DDHD2 gene (8p11.23) encoding phospholipase DDHD2.