

DISEASE:
Autosomal recessive spastic paraplegia type 45

NAME:	Autosomal recessive spastic paraplegia type 45
DESCRIPTION:	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.
ORPHACODE:	320396
SYNOMYS:	Autosomal recessive spastic paraplegia type 65 SPG45 SPG65
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	NT5C2
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