DISEASE:
Autosomal recessive spastic paraplegia type 20

NAME:	Autosomal recessive spastic paraplegia type 20
DESCRIPTION:	Autosomal recessive spastic paraplegia type 20 (SPG20) is a type of complex hereditary spastic paraplegia characterized by an onset in infancy of progressive spastic paraparesis associated with distal amyotrophy, psuedobulbar palsy, motor and cognitive delays, mild cerebellar signs (dysarthria, dysdiadochokinesia, mild intention tremor), short stature and subtle skeletal abnormalities (pes cavus, mild talipes equinovarus, kyphoscoliosis). SPG20 is due to mutations in the SPG20 gene (13q13.1), which encodes the protein spartin.
ORPHACODE:	101000
SYNONYMS:	Childhood-onset spastic paraparesis-distal muscle wasting syndrome SPG20 Troyer syndrome
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	SPART
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/3720

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