

Full name:	Spastic Paraplegia (89 genes) - IPG
Version number:	V4
Laboratory:	<u>Centre de Génétique-Institut de Pathologie et de Génétique (IPG)</u>
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Related Diseases

- 3C syndrome
- Allan-Herndon-Dudley syndrome
- Ataxia-hypogonadism-choroidal dystrophy syndrome
- Autosomal dominant spastic paraplegia type 10
- Autosomal dominant spastic paraplegia type 12
- Autosomal dominant spastic paraplegia type 13
- Autosomal dominant spastic paraplegia type 17
- Autosomal dominant spastic paraplegia type 3
- Autosomal dominant spastic paraplegia type 31
- Autosomal dominant spastic paraplegia type 4
- Autosomal dominant spastic paraplegia type 42
- Autosomal dominant spastic paraplegia type 6
- Autosomal dominant spastic paraplegia type 8
- Autosomal recessive cerebellar ataxia with late-onset spasticity
- Autosomal recessive spastic ataxia of Charlevoix-Saguenay
- Autosomal recessive spastic paraplegia type 11
- Autosomal recessive spastic paraplegia type 15
- Autosomal recessive spastic paraplegia type 20
- Autosomal recessive spastic paraplegia type 21
- Autosomal recessive spastic paraplegia type 26
- Autosomal recessive spastic paraplegia type 28
- Autosomal recessive spastic paraplegia type 35
- Autosomal recessive spastic paraplegia type 39

- [Autosomal recessive spastic paraplegia type 43](#)
- [Autosomal recessive spastic paraplegia type 44](#)
- [Autosomal recessive spastic paraplegia type 45](#)
- [Autosomal recessive spastic paraplegia type 46](#)
- [Autosomal recessive spastic paraplegia type 48](#)
- [Autosomal recessive spastic paraplegia type 53](#)
- [Autosomal recessive spastic paraplegia type 54](#)
- [Autosomal recessive spastic paraplegia type 55](#)
- [Autosomal recessive spastic paraplegia type 56](#)
- [Autosomal recessive spastic paraplegia type 57](#)
- [Autosomal recessive spastic paraplegia type 59](#)
- [Autosomal recessive spastic paraplegia type 5A](#)
- [Autosomal recessive spastic paraplegia type 60](#)
- [Autosomal recessive spastic paraplegia type 61](#)
- [Autosomal recessive spastic paraplegia type 62](#)
- [Autosomal recessive spastic paraplegia type 63](#)
- [Autosomal recessive spastic paraplegia type 64](#)
- [Autosomal recessive spastic paraplegia type 66](#)
- [Autosomal recessive spastic paraplegia type 67](#)
- [Autosomal recessive spastic paraplegia type 69](#)
- [Autosomal recessive spastic paraplegia type 70](#)
- [Autosomal recessive spastic paraplegia type 71](#)
- [Autosomal recessive spastic paraplegia type 75](#)
- [Autosomal spastic paraplegia type 18](#)
- [Autosomal spastic paraplegia type 30](#)
- [Autosomal spastic paraplegia type 58](#)
- [Autosomal spastic paraplegia type 72](#)
- [BICD2-related autosomal dominant childhood-onset proximal spinal muscular atrophy](#)
- [Cerebellar ataxia-hypogonadism syndrome](#)
- [Fatty acid hydroxylase-associated neurodegeneration](#)
- [Hereditary sensory and autonomic neuropathy due to TECPR2 mutation](#)
- [Hereditary sensory and autonomic neuropathy type 1](#)
- [Infantile-onset ascending hereditary spastic paralysis](#)
- [Inherited congenital spastic tetraplegia](#)
- [Juvenile amyotrophic lateral sclerosis](#)
- [Juvenile primary lateral sclerosis](#)

- Mutilating hereditary sensory neuropathy with spastic paraplegia
- Oculodentodigital dysplasia
- Pontocerebellar hypoplasia type 9
- Recessive intellectual disability-motor dysfunction-multiple joint contractures syndrome
- Severe intellectual disability and progressive spastic paraplegia
- Spastic paraplegia type 2
- Spastic paraplegia type 7
- Spastic paraplegia-Paget disease of bone syndrome
- Spastic paraplegia-optic atrophy-neuropathy syndrome
- Spinocerebellar ataxia with axonal neuropathy type 2
- X-linked complicated spastic paraplegia type 1
- X-linked spastic paraplegia type 34

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ADAR</u>	100.00	1	NM_001111.5
<u>AFG3L2</u>	100.00	1	NM_006796.3
<u>ALDH18A1</u>	100.00	1	NM_002860.4
<u>ALDH3A2</u>	100.00	1	NM_000382.3
<u>ALS2</u>	100.00	1	NM_020919.4
<u>AMPD2</u>	100.00	1	NM_001368809.2
<u>AP4B1</u>	100.00	1	NM_001253852.3
<u>AP4E1</u>	100.00	1	NM_007347.5
<u>AP4M1</u>	100.00	1	NM_004722.4
<u>AP4S1</u>	100.00	1	NM_001128126.3
<u>AP5Z1</u>	100.00	1	NM_014855.3
<u>ARL6IP1</u>	100.00	1	NM_015161.3

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ARSI</u>	100.00	1	NM_001012301.4
<u>ATL1</u>	100.00	1	NM_015915.5
<u>ATXN1</u>	100.00	1	NM_000332.3
<u>B4GALNT1</u>	100.00	1	NM_001478.5
<u>BICD2</u>	100.00	1	NM_001003800.2
<u>BSCL2</u>	100.00	1	NM_001122955.3
<u>C19ORF12</u>	100.00	1	NM_031448.6
<u>CAPN1</u>	100.00	1	NM_005186.4
<u>CCT5</u>	100.00	1	NM_012073.5
<u>COA8</u>	100.00	1	NM_001370595.1
<u>CPT1C</u>	100.00	1	NM_001199753.1
<u>CYP27A1</u>	100.00	1	NM_000784.4
<u>CYP2U1</u>	100.00	1	NM_183075.3
<u>CYP7B1</u>	100.00	1	NM_004820.5
<u>DDHD1</u>	100.00	1	NM_001160148.2
<u>DDHD2</u>	100.00	1	NM_015214.3
<u>DSTYK</u>	100.00	1	NM_015375.3
<u>ENTPD1</u>	100.00	1	NM_001776.6
<u>ERLIN1</u>	100.00	1	NM_006459.4
<u>ERLIN2</u>	100.00	1	NM_007175.8

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>FA2H</u>	100.00	1	NM_024306.5
<u>FARS2</u>	100.00	1	NM_006567.5
<u>FLRT1</u>	100.00	1	NM_013280.4
<u>GAD1</u>	100.00	1	NM_000817.3
<u>GBA2</u>	100.00	1	NM_020944.3
<u>GFAP</u>	100.00	1	NM_002055.5
<u>GJA1</u>	100.00	1	NM_000165.5
<u>GJC2</u>	100.00	1	NM_020435.4
<u>GLRX5</u>	100.00	1	NM_016417.3
<u>HACE1</u>	100.00	1	NM_020771.4
<u>HSPD1</u>	100.00	1	NM_002156.5
<u>IBA57</u>	100.00	1	NM_001010867.4
<u>ITPR1</u>	100.00	1	NM_001168272.1
<u>KIDINS220</u>	100.00	1	NM_020738.4
<u>KIF1A</u>	100.00	1	NM_001244008.1
<u>KIF1C</u>	100.00	1	NM_006612.6
<u>KIF5A</u>	100.00	1	NM_004984.4
<u>KLC2</u>	100.00	1	NM_001134775.1
<u>KLC4</u>	100.00	1	NM_201521.3
<u>L1CAM</u>	100.00	1	NM_001278116.2

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>MAG</u>	100.00	1	NM_002361.4
<u>MARS1</u>	100.00	1	NM_004990.4
<u>MTRFR</u>	100.00	1	NM_152269.5
<u>NIPA1</u>	100.00	1	NM_144599.5
<u>NT5C2</u>	100.00	1	NM_001351169.2
<u>PCYT2</u>	100.00	1	NM_002861.5
<u>PGAP1</u>	100.00	1	NM_024989.4
<u>PLP1</u>	100.00	1	NM_000533.5
<u>PNPLA6</u>	100.00	1	NM_001166111.2
<u>RAB18</u>	100.00	1	NM_021252.5
<u>RAB3GAP1</u>	100.00	1	NM_012233.3
<u>RAB3GAP2</u>	100.00	1	NM_012414.4
<u>REEP1</u>	100.00	1	NM_001371279.1
<u>REEP2</u>	100.00	1	NM_001271803.2
<u>RTN2</u>	100.00	1	NM_005619.5
<u>SACS</u>	100.00	1	NM_014363.6
<u>SETX</u>	100.00	1	NM_015046.7
<u>SLC16A2</u>	100.00	1	NM_006517.5
<u>SLC33A1</u>	100.00	1	NM_004733.4
<u>SPART</u>	100.00	1	NM_015087.5

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>SPAST</u>	100.00	1	NM_014946.3
<u>SPG11</u>	100.00	1	NM_025137.4
<u>SPG21</u>	100.00	1	NM_016630.7
<u>SPG7</u>	100.00	1	NM_003119.4
<u>TBC1D20</u>	100.00	1	NM_144628.4
<u>TECPR2</u>	100.00	1	NM_014844.5
<u>TFG</u>	100.00	1	NM_006070.6
<u>UBAP1</u>	100.00	1	NM_016525.5
<u>UCHL1</u>	100.00	1	NM_004181.5
<u>USP8</u>	100.00	1	NM_005154.5
<u>VCP</u>	100.00	1	NM_007126.5
<u>VPS37A</u>	100.00	1	NM_152415.3
<u>WASHC5</u>	100.00	1	NM_014846.4
<u>WDR48</u>	100.00	1	NM_020839.4
<u>ZFR</u>	100.00	1	NM_016107.5
<u>ZFYVE26</u>	100.00	1	NM_015346.4
<u>ZFYVE27</u>	100.00	1	NM_001002261.3