



Liste des gènes étudiés en diagnostic moléculaire avec le panel spécifique aux paraplégies spastiques héréditaires

Gène	N° SPG (ou syndrome associé)	Transmission	Références (selon La Giudice <i>et al.</i> , Exp Neurol. 2014)	N° Gène OMIM
ABCD1	<i>aucun n° SPG</i>	X	Mosser J <i>et al.</i> , Nature 1993	300371
ALDH18A1	SPG9	AD et AR	Coutelier M <i>et al.</i> , Brain 2015	138250
ALS2	<i>aucun n° SPG</i>	AR	Wakil SM <i>et al.</i> , Gene 2014	606352
AMPD2	SPG63	AR	Novarino G <i>et al.</i> , Science 2014	102771
AP4B1	SPG47	AR	Bauer P <i>et al.</i> , Neurogenetics 2012	607245
AP4E1	SPG51	AR	Abou Jamra R <i>et al.</i> , Am. J. Hum. Genet. 2011	607244
AP4M1	SPG50	AR	Abou Jamra R <i>et al.</i> , Am. J. Hum. Genet. 2011	602296
AP4S1	SPG52	AR	Abou Jamra R <i>et al.</i> , Am. J. Hum. Genet. 2011	607243
AP5Z1	SPG48	AR	Slabicki M <i>et al.</i> , PLoS Biol. 2010	613653
ARL6IP1	SPG61	AR	Novarino G <i>et al.</i> , Science 2014	607669
ARSI	SPG66	AR	Novarino G <i>et al.</i> , Science 2014	610009
ATL1	SPG3A	AD	Zhao X <i>et al.</i> , Nature Genet. 2001	606439
ATP13A2	SPG78	AR	Estrada-Cuzcano A <i>et al.</i> , Brain 2017	610513
B4GALNT1	SPG26	AR	Boukhris A <i>et al.</i> , Am. J. Hum. Genet. 2013	601873
BICD2	<i>aucun n° SPG</i>	AD	Novarino G <i>et al.</i> , Science 2014	609797
BSC12	SPG17	AD	Windpassinger C <i>et al.</i> , Nature Genet. 2004	606158
C12orf65	SPG55	AR	Shimazaki H <i>et al.</i> , J. Med. Genet. 2012	613541
C19orf12	SPG43	AR	Landoure G <i>et al.</i> , Hum. Mutat. 2013	614297
CAPN1	SPG76	AR	Gan-Or Z <i>et al.</i> , Am. J. Hum. Genet. 2016	114220
CCT5	<i>aucun n° SPG</i>	AR	Bouhouche A <i>et al.</i> , J. Med. Genet. 2006	610150
CPT1C	SPG73	AD	Rinaldi C <i>et al.</i> , JAMA Neurol. 2015	608846
CYP2U1	SPG56	AR	Tesson C <i>et al.</i> , Am. J. Hum. Genet. 2012	610670
CYP7B1	SPG5A	AR	Tsaousidou M <i>et al.</i> , Am. J. Hum. Genet. 2008	603711
DDHD1	SPG28	AR	Tesson C <i>et al.</i> , Am. J. Hum. Genet. 2012	614603
DDHD2	SPG54	AR	Schuurs-Hoeijmakers JHM <i>et al.</i> , Am. J. Hum. Genet. 2012	615003
ENTPD1	SPG64	AR	Novarino G <i>et al.</i> , Science 2014	601752
ERLIN1	SPG62	AR	Novarino G <i>et al.</i> , Science 2014	611604
ERLIN2	SPG18	AR	Alazami A <i>et al.</i> , Neurogenetics 2011	611605
FA2H	SPG35	AR	Dick KJ <i>et al.</i> , Hum. Mutat. 2010	611026
FBXO7	Pallido-pyramidal syndrome	AR	Di Fonzo A <i>et al.</i> , Neurology 2009	605648
FLRT1	SPG68	AR	Novarino G <i>et al.</i> , Science 2014	604806
GAD1	<i>aucun n° SPG</i>	AR	Lynex CN <i>et al.</i> , BMC Neurol. 2004	605363
GBA2	SPG46	AR	Martin E <i>et al.</i> , Am. J. Hum. Genet. 2013.	609471
GJA1	Pleiotropic Phenotype of Oculodentodigital Dysplasia	AR	Paznekas WA <i>et al.</i> , Am. J. Hum. Genet. 2003	121014
GJC2	SPG44	AR	Orthmann-Murphy JL <i>et al.</i> , Brain 2009	608803
HSPD1	SPG13	AD	Hansen JJ <i>et al.</i> , Am. J. Hum. Genet. 2002	118190
KIAA0196	SPG8	AD	Valdmanis PN <i>et al.</i> , Am. J. Hum. Genet. 2007	610657
KIF1A	SPG30	AD et AR	Erich Y <i>et al.</i> , Genome Res. 2011	601255
KIF1C	SPG58	AR	Novarino G <i>et al.</i> , Science 2014	603060
KIF5A	SPG10	AD	Reid E <i>et al.</i> , Am. J. Hum. Genet. 2002	602821
L1CAM	SPG1	X	Jouet M <i>et al.</i> , Nature Genet. 1994	308840
LYST	<i>aucun n° SPG</i>	AR	Shimazaki H <i>et al.</i> , Neurogenetics 2014	606897
MAG	<i>aucun n° SPG</i>	AR	Novarino G <i>et al.</i> , Science 2014	159460
MARS	SPG70	AR	Novarino G <i>et al.</i> , Science 2014	156560
NIPA1	SPG6	AD	Rainier S <i>et al.</i> , Am. J. Hum. Genet. 2003	608145
NT5C2	SPG65	AR	Novarino G <i>et al.</i> , Science 2014	600417
PGAP1	SPG67	AR	Novarino G <i>et al.</i> , Science 2014	611655
PLP1	SPG2	X	Saugier-veber P <i>et al.</i> , Nature Genet. 1994	300401
PNPLA6	SPG39	AR	Rainier S <i>et al.</i> , Am. J. Hum. Genet. 2008	603197
PSEN1	<i>aucun n° SPG</i>	AD	Crook R <i>et al.</i> , Nature Medicine 1998	104311
RAB3GAP2	SPG69	AR	Novarino G <i>et al.</i> , Science 2014	609275
REEP1	SPG31	AD	Zuchner S <i>et al.</i> , Am. J. Hum. Genet. 2006	609139
REEP2	SPG72	AD et AR	Esteves T <i>et al.</i> , Am. J. Hum. Genet. 2014	609347
RTN2	SPG12	AD	Montenegro G <i>et al.</i> , J. Clin. Invest. 2012	603183
SACS	ARSACS	AR	Engert JC <i>et al.</i> , Nature Genet. 2000	604490
SAMHD1	Aicardi-Goutières syndrome 5 (AGS5)	AR	Rice G <i>et al.</i> , Nature Genet. 2009	606754
SETX	Juvenile Amyotrophic Lateral Sclerosis 4	AD et AR	Chen YZ <i>et al.</i> , Am. J. Hum. Genet. 2004	608465
SLC16A2	SPG22	X	Schwartz CE <i>et al.</i> , Am. J. Hum. Genet. 2005	300095
SLC33A1	SPG42	AD	Lin P <i>et al.</i> , Am. J. Hum. Genet. 2008	603690
SPAST	SPG4	AD	Hazan J <i>et al.</i> , Nature Genet. 1999	604277
SPG11	SPG11	AR	Stevanin G <i>et al.</i> , Nature Genet. 2007	610844
SPG20	SPG20	AR	Patel H <i>et al.</i> , Nature Genet. 2002	607111
SPG21	SPG21	AR	Simpson MA <i>et al.</i> , Am. J. Hum. Genet. 2003	248900
SPG7	SPG7	AD et AR	Casari G <i>et al.</i> , Cell 1998	602783
TECP2	SPG49	AR	Oz-Levi D <i>et al.</i> , Am. J. Hum. Genet. 2012	615000
TFG	SPG57	AR	Beetz C <i>et al.</i> , Proc. Nat. Acad. Sci. 2013	602498
USP8	SPG59	AR	Novarino G <i>et al.</i> , Science 2014	603158
VCP	<i>aucun n° SPG</i>	AD	de Bot ST <i>et al.</i> , Brain 2012	610123
VPS37A	SPG53	AR	Zivony-Elboum Y <i>et al.</i> , J. Med. Genet. 2012	609927
WDR48	SPG60	AR	Novarino G <i>et al.</i> , Science 2014	612167
ZFR	SPG71	AR	Novarino G <i>et al.</i> , Science 2014	615635
ZFYVE26	SPG15	AR	Hanein S <i>et al.</i> , Am. J. Hum. Genet. 2008	612012

Transmission : AD : autosomique dominant ; AR : autosomique récessive ; X : liée au chromosome X.