



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

Gène	N° SPG (ou syndrome associé)	Références (selon Lo Giudice <i>et al.</i> , Exp Neurol. 2014)	N° Gène OMIM	N° LRG (transcript de référence associé)
ALS2	<i>aucun n° SPG</i>	Wakil SM <i>et al.</i> , Gene 2014	606352	[‡] LRG_654 (NM_020919.3)
AMPD2	SPG63	Novarino G <i>et al.</i> , Science 2014	102771	nd
AP4B1	SPG47	Bauer P <i>et al.</i> , Neurogenetics 2012	607245	nd
AP4E1	SPG51	Abou Jamra R <i>et al.</i> , Am. J. Hum. Genet. 2011	607244	[‡] LRG_732 (NM_007347.4)
AP4M1	SPG50	Abou Jamra R <i>et al.</i> , Am. J. Hum. Genet. 2011	602296	nd
AP4S1	SPG52	Abou Jamra R <i>et al.</i> , Am. J. Hum. Genet. 2011	607243	nd
AP5Z1	SPG48	Slabicki M <i>et al.</i> , PLoS Biol. 2010	613653	nd
ARL6IP1	SPG61	Novarino G <i>et al.</i> , Science 2014	607669	nd
ARSI	SPG66	Novarino G <i>et al.</i> , Science 2014	610009	nd
ATL1	SPG3A	Zhao X <i>et al.</i> , Nature Genet. 2001	606439	[‡] LRG_360 (*multiple)
B4GALNT1	SPG26	Boukhris A <i>et al.</i> , Am. J. Hum. Genet. 2013	601873	nd
BICD2	<i>aucun n° SPG</i>	Novarino G <i>et al.</i> , Science 2014	609797	nd
BSC12	SPG17	Windpassinger C <i>et al.</i> , Nature Genet. 2004	606158	[‡] LRG_235 (NM_001122955.3)
C12orf65	SPG55	Shimazaki H <i>et al.</i> , J. Med. Genet. 2012	613541	nd
C19orf12	SPG43	Landouze G <i>et al.</i> , Hum. Mutat. 2013	614297	nd
CCT5	<i>aucun n° SPG</i>	Bouhouche A <i>et al.</i> , J. Med. Genet. 2006	610150	LRG_361 (NM_012073.3)
CYP2U1	SPG56	Tesson C <i>et al.</i> , Am. J. Hum. Genet. 2012	610670	nd
CYP7B1	SPG5A	Tsaousidou M <i>et al.</i> , Am. J. Hum. Genet. 2008	603711	nd
DDHD1	SPG28	Tesson C <i>et al.</i> , Am. J. Hum. Genet. 2012	614603	nd
DDHD2	SPG54	Schuurs-Hoeijmakers JHM <i>et al.</i> , Am. J. Hum. Genet. 2012	615003	nd
ENTPD1	SPG64	Novarino G <i>et al.</i> , Science 2014	601752	nd
ERLIN1	SPG62	Novarino G <i>et al.</i> , Science 2014	611604	nd
ERLIN2	SPG18	Alazami A <i>et al.</i> , Neurogenetics 2011	611605	nd
FA2H	SPG35	Dick KJ <i>et al.</i> , Hum. Mutat. 2010	611026	nd
FBXO7	Pallido-pyramidal syndrome	Di Fonzo A <i>et al.</i> , Neurology 2009	605648	nd
FLRT1	SPG68	Novarino G <i>et al.</i> , Science 2014	604806	nd
GAD1	<i>aucun n° SPG</i>	Lynex CN <i>et al.</i> , BMC Neurol. 2004	605363	nd
GBA2	SPG46	Martin E <i>et al.</i> , Am. J. Hum. Genet. 2013.	609471	nd
GJA1	Pleiotropic Phenotype of Oculodentodigital Dysplasia	Paznekas WA <i>et al.</i> , Am. J. Hum. Genet. 2003	121014	nd
GJC2	SPG44	Orthmann-Murphy JL <i>et al.</i> , Brain 2009	608803	nd
HSPD1	SPG13	Hansen JJ <i>et al.</i> , Am. J. Hum. Genet. 2002	118190	nd
KIAA0196	SPG8	Valdmanis PN <i>et al.</i> , Am. J. Hum. Genet. 2007	610657	nd
KIF1A	SPG30	Erlich Y <i>et al.</i> , Genome Res. 2011	601255	[‡] LRG_367 (*multiple)
KIF1C	SPG58	Novarino G <i>et al.</i> , Science 2014	603060	nd
KIF5A	SPG10	Reid E <i>et al.</i> , Am. J. Hum. Genet. 2002	602821	nd
L1CAM	SPG1	Jouet M <i>et al.</i> , Nature Genet. 1994	308840	[‡] LRG_14 (NM_000425.3)
MAG	<i>aucun n° SPG</i>	Novarino G <i>et al.</i> , Science 2014	159460	nd
MARS	SPG70	Novarino G <i>et al.</i> , Science 2014	156560	nd
NIPA1	SPG6	Rainier S <i>et al.</i> , Am. J. Hum. Genet. 2003	608145	nd
NT5C2	SPG65	Novarino G <i>et al.</i> , Science 2014	600417	nd
PGAP1	SPG67	Novarino G <i>et al.</i> , Science 2014	611655	nd
PLP1	SPG2	Saugier-Verber P <i>et al.</i> , Nature Genet. 1994	300401	nd
PNPLA6	SPG39	Rainier S <i>et al.</i> , Am. J. Hum. Genet. 2008	603197	nd
RAB3GAP2	SPG69	Novarino G <i>et al.</i> , Science 2014	609275	nd
REEP1	SPG31	Zuchner S <i>et al.</i> , Am. J. Hum. Genet. 2006	609139	[‡] LRG_713 (*multiple)
REEP2	SPG72	Esteves T <i>et al.</i> , Am. J. Hum. Genet. 2014	609347	nd
RTN2	SPG12	Montenegro G <i>et al.</i> , J. Clin. Invest. 2012	603183	nd
SACS	ARSACS	Engert JC <i>et al.</i> , Nature Genet. 2000	604490	nd
SAMHD1	Aicardi-Goutières syndrome 5 (AGS5)	Rice G <i>et al.</i> , Nature Genet. 2009	606754	LRG_281 (NM_015474.3)
SETX	Juvenile Amyotrophic Lateral Sclerosis 4	Chen YZ <i>et al.</i> , Am. J. Hum. Genet. 2004	608465	LRG_268 (NM_015046.5)
SLC16A2	SPG22	Schwartz CE <i>et al.</i> , Am. J. Hum. Genet. 2005	300095	nd
SLC33A1	SPG42	Lin P <i>et al.</i> , Am. J. Hum. Genet. 2008	603690	nd
SPAST	SPG4	Hazan J <i>et al.</i> , Nature Genet. 1999	604277	[‡] LRG_714 (NM_014946.3)
SPG11	SPG11	Stevanin G <i>et al.</i> , Nature Genet. 2007	610844	nd
SPG20	SPG20	Patel H <i>et al.</i> , Nature Genet. 2002	607111	nd
SPG21	SPG21	Simpson MA <i>et al.</i> , Am. J. Hum. Genet. 2003	248900	nd
SPG7	SPG7	Casari G <i>et al.</i> , Cell 1998	602783	nd
TECPR2	SPG49	Oz-Levi D <i>et al.</i> , Am. J. Hum. Genet. 2012	615000	nd
TFG	SPG57	Beetz C <i>et al.</i> , Proc. Nat. Acad. Sci. 2013	602498	nd
USP8	SPG59	Novarino G <i>et al.</i> , Science 2014	603158	nd
VCP	<i>aucun n° SPG</i>	de Bot ST <i>et al.</i> , Brain 2012	601023	LRG_657 (NM_007126.3)
VPS37A	SPG53	Zivony-Elboum Y <i>et al.</i> , J. Med. Genet. 2012	609927	nd
WDR48	SPG60	Novarino G <i>et al.</i> , Science 2014	612167	nd
ZFR	SPG71	Novarino G <i>et al.</i> , Science 2014	615635	nd
ZFYVE26	SPG15	Hanein S <i>et al.</i> , Am. J. Hum. Genet. 2008	612012	nd

[‡]En attente d'approbation dans la base de données LRG. *Plusieurs transcrits de référence disponibles dans la base de données LRG. Site internet de la base de données LRG (Locus Reference Genomic) : <http://www.lrg-sequence.org/>. Site internet de la base de données OMIM (Online Mendelian Inheritance in Man) : <http://www.ncbi.nlm.nih.gov/omim>.