



Liste des gènes étudiés pour le diagnostic moléculaire de la sclérose latérale amyotrophique avec la technique de Next Generation Sequencing (NGS)

Gène	Description	N°SLA	Locus	Mode de transmission	Référence	Phenotype MIM number	Pathologie OMIM	Transcrit utilisé
ALS2	Alsin	ALS2	2q33.1	AR	Yang Y et al, 2001 [PMID:11586297]	205100	Amyotrophic lateral sclerosis 2, juvenile	NM_020919.3
ANG	Angiogenin	ALS9	14q11.2	AD	van Es MA et al, 2009 [PMID:19153377]	611895	Amyotrophic lateral sclerosis 9	NM_001145.4
CHMP2B	CHMP family, member 2b	ALS17	3p11.2	AD	Skibinski G et al, 2008 [PMID:16041373]	614696	Amyotrophic lateral sclerosis 17	NM_014043.3
DCTN1	Dynactin subunit 1	-	2p13.1	AD	Puls I et al, 2003 [PMID:12627231]	607641	distal hereditary motor neuropathy type VIIB	NM_004082.4
FIG4	Polyphosphoinositide phosphatase	ALS11	6q21	AD	Chow CY et al, 2009 [PMID:19118816]	612577	Amyotrophic lateral sclerosis 11	NM_014845.5
FUS	RNA-binding protein FUS	ALS6	16p11.2	AD	Kwiatkowski TJ Jr et al, 2009 [PMID:19251627]	608030	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia	NM_004960.3
OPTN	Optineurin	ALS12	10p13	AR	Maruyama et al, 2010 [PMID:20428114]	613435	Amyotrophic lateral sclerosis 12	NM_001008211.1
SETX	Probable helicase senataxin	ALS4	9q34.13	AD	Chen YZ et al, 2004 [PMID:15106121]	602433	Amyotrophic lateral sclerosis 4, juvenile	NM_015046.5
SOD1	Superoxide dismutase (Cu-Zn)	ALS1	21q22.11	AD	Rosen DR et al, 1993 [PMID:8446170]	105400	Amyotrophic lateral sclerosis 1	NM_000454.4
SPG20	Spartin	SPG20	13q13.3	AR	Patel et al, 2002 [PMID:12134148]	275900	Troyer syndrome	NM_001142296.1
SQSTM1	Sequestosome 1	-	5q35.3	AD	Fecto F et al, 2011 [PMID:22084127]	616437	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3	NM_003900.4
TARDBP	TAR DNA-binding protein 43	ALS10	1p36.22	AD	Sreedharan J et al, 2008 [PMID:18309045]	612069	Amyotrophic lateral sclerosis 10, with or without FTD	NM_007375.3
TBK1	TANK binding kinase	FTDALS4	12q14.2	AD	Cirulli ET et al, 2015 [PMID:25700176], Freischmidt A et al, 2015 [PMID:25803835]	616439	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4	NM_013254.3
UBQLN2	Ubiquilin-2	ALS15	Xp11.21	XL	Deng HX et al, 2011 [PMID:21857683]	300857	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia	NM_013444.3
VAPB	Vesicle-associated membrane protein-associated protein B/C	ALS8	20q13.32	AD	Nishimura AL et al, 2004 [PMID:15372378]	608627	Amyotrophic lateral sclerosis 8	NM_004738.4
VCP	Transitional endoplasmic reticulum ATPase	ALS14	9p13.3	AD	Johnson JO et al, 2010 [PMID:21145000]	613954	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia	NM_007126.3