



Liste des gènes étudiés en diagnostic moléculaire de la sclérose latérale amyotrophique avec le panel NGS

Gène	N°ALS	Mode de transmission	Pathologie OMIM	Référence
ANXA11	ALS23	AD	Amyotrophic lateral sclerosis 23	Smith et al., 2017 [PMID: 28469040]
ALS2	ALS2	AR	Amyotrophic lateral sclerosis 2, juvenile	Yang Y et al., 2001 [PMID:11586297]
ANG	ALS9	AD	Amyotrophic lateral sclerosis 9	van Es MA et al., 2009 [PMID:19153377]
CHCHD10	FTDALS2	AD	Frontotemporal dementia and/or amyotrophic lateral sclerosis 2	Bannwarth et al., 2014 [PMID: 24934289]
CHMP2B	ALS17	AD	Amyotrophic lateral sclerosis 17	Skibinski G et al., 2008 [PMID:16041373]
DCTN1	-	AD	distal hereditary motor neuronopathy type VIIB	Puls I et al., 2003 [PMID:12627231]
FIG4	ALS11	AD	Amyotrophic lateral sclerosis 11	Chow CY et al., 2009 [PMID:19118816]
FUS	ALS6	AD	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia	Kwiatkowski TJ Jr et al., 2009 [PMID:19251627]
KIF5A	ALS25	AD	Amyotrophic lateral sclerosis, susceptibility to, 25	Nicolas et al., 2018 [PMID: 29566793]
MATR3	ALS21	AD	Amyotrophic lateral sclerosis 21	Johnson et al., 2014 [PMID: 24686783]
NEK1	ALS24	AD	Amyotrophic lateral sclerosis, susceptibility to, 24	Brenner et al., 2016 [PMID: 26945885]
OPTN	ALS12	AR	Amyotrophic lateral sclerosis 12	Maruyama et al., 2010 [PMID:20428114]
SOD1	ALS1	AD	Amyotrophic lateral sclerosis 1	Rosen DR et al., 1993 [PMID:8446170]
SQSTM1	FTDALS3	AD	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3	Fecto F et al., 2011 [PMID:22084127]
TARDBP	ALS10	AD	Amyotrophic lateral sclerosis 10, with or without FTD	Sreedharan J et al., 2008 [PMID:18309045]
TBK1	FTDALS4	AD	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4	Cirulli ET et al., 2015 [PMID:25700176], Freischmidt A et al., 2015 [PMID:25803835]
TUBA4A	ALS22	AD	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia	Smith et al., 2014 [PMID: 25374358]
UBQLN2	ALS15	XL	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia	Deng HX et al., 2011 [PMID:21857683]
VAPB	ALS8	AD	Amyotrophic lateral sclerosis 8	Nishimura AL et al., 2004 [PMID:15372378]
VCP	ALS14	AD	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia	Johnson JO et al., 2010 [PMID:21145000]