



Liste des gènes étudiés en diagnostic moléculaire des dégénérescences lobaires fronto-temporales avec le panel NGS

Gène	Mode de transmission	Pathologie OMIM	Référence
CHMP2B	AD	Frontotemporal dementia and/or amyotrophic lateral sclerosis 7	Gydesen et al., 2002; Skibinski et al., 2005 ; Parkinson et al., 2006
CSF1R	AD	Leukoencephalopathy diffuse hereditary with spheroids	Rademakers et al., 2011
DCTN1	AD	Perry syndrome	Farrer et al., 2009
FUS	AD	Amyotrophic lateral sclerosis 6 with or without frontotemporal dementia	Lattante et al., 2013
GRN	AD	Frontotemporal lobar degeneration with ubiquitin-positive inclusions	Baker et al., 2006 ; Cruts et al., 2006
HNRNPA2B1	AD	Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2	Waggoner et al., 2002
MAPT / TAU	AD	Dementia frontotemporal with or without parkinsonism ; Supranuclear palsy progressive	Hutton et al., 1998
SQSTM1	AD	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3; Paget disease of bone 3	Kurihara et al., 2007; Fecto et al., 2011
TARDBP	AD	Amyotrophic lateral sclerosis 10 with or without FTD	Gitcho et al., 2009; Lattante et al., 2013
TBK1	AD	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4	Cirulli et al., 2015; Pottier et al., 2015
TREM2	AR	Nasu-Hakola disease	Klunemann et al., 2005
TUBA4A	AD	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia	Smith et al. (2014)
UBQLN2	XD	Amyotrophic lateral sclerosis 15 with or without frontotemporal dementia	Deng et al., 2011; Fahed et al., 2014
VCP	AD	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1	Mehta et al., 2013