



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

Gène	Mode de transmission	Pathologie OMIM	Référence
<i>CHMP2B</i>	AD	Amyotrophic lateral sclerosis 17 ; Dementia familial non specific	Gydesen et al., 2002, Skibinski et al., 2005 ; Parkinson et al., 2006
<i>CSF1R</i>	AD	Leukoencephalopathy diffuse hereditary with spheroids	Rademakers et al., 2011
<i>DCTN1</i>	AD	Perry syndrome	Farrer et al., 2009
<i>FUS</i>	AD	Amyotrophic lateral sclerosis 6 with or without frontotemporal dementia	Lattante et al., 2013
<i>GRN</i>	AD	Frontotemporal lobar degeneration with ubiquitin-positive inclusions	Baker et al., 2006 ; Cruts et al., 2006
<i>HNRNPA2B1</i>	AD	Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2	Waggoner et al., 2002
<i>MAPT / TAU</i>	AD	Dementia frontotemporal with or without parkinsonism ; Supranuclear palsy progressive	Hutton et al., 1998
<i>SQSTM1</i>	AD	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3; Paget disease of bone 3	Kurihara et al., 2007; Fecto et al., 2011
<i>TARDBP</i>	AD	Amyotrophic lateral sclerosis 10 with or without FTD	Gitcho et al., 2009; Lattante et al., 2013
<i>TBK1</i>	AD	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4	Cirulli et al., 2015; Pottier et al., 2015
<i>TREM2</i>	AR	Nasu-Hakola disease	Klunemann et al., 2005
<i>UBQLN2</i>	XD	Amyotrophic lateral sclerosis 15 with or without frontotemporal dementia	Deng et al., 2011; Fahed et al., 2014
<i>VCP</i>	AD	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1	Mehta et al., 2013