

Table S1. Summary of excluded studies.

Author	Year	Journal	Reason for exclusion
Nakano R[1]	1994	Biochem Biophys Res Commun	Unclear diagnostic criteria
Juneja T[2]	1997	Neurology	Unclear diagnostic criteria
Shaw PJ[3]	1997	Neuroreport	Unclear diagnostic criteria
Luquin N[4]	2009	Neuromuscular Disorders	Unclear diagnostic criteria
Aoki M[5]	1993	Nat Genet	Same patient group as Aoki M, 1998
Deng HX[6]	1993	Science	Same patient group as Deng XH, 1995
Rosen DR[7]	1993	Nature	Same patient group as Cudkowicz ME, 1997
Jones CT[8]	1994	Mol Cell Probes	Same patient group as Jones CT, 1995
Jones CT[9]	1994	Hum Mol Genet	Same patient group as Jones CT, 1995
Abe K[10]	1996	J Neurol Sci	Same patient group as Aoki M, 1998
Andersen PM[11]	1995	Nat Genet	Same patient group as Andersen PM, 1997
Andersen PM[12]	1996	Brain	Same patient group as Andersen PM, 1997
Enayat ZE[13]	1995	Hum Mol Genet	Same patient group as Orrell RW, 1999
Orrell RW[14]	1997	Neurology	Same patient group as Orrell RW, 1999
Gellera C[15]	2001	Neuromuscul Disord	Same patient group as Gellera C, 2001
Restagno G[16]	2005	Amyotroph Lateral Scler Other Motor Neuron Disord	Same patient group as Chio A, 2008
Lemmens R[17]	2009	J Neurol Neurosurg Psychiatry	Same patient group as Debray S, 2013
Damme PV[18]	2010	Eur J Neurol	Same patient group as Debray S,2013
Groen EJ[19]	2010	Arch Neurol	Same patient group as van Blitterswijk M, 2012
Millicamps S[20]	2010	J Med Genet	Same patient group as Millicamps S,2012
van Es MA[21]	2010	J Neurol Neurosurg Psychiatry	Same patient group as van Blitterswijk M,2012
Waibel S[22]	2010	Neurology	Same patient group as Waibel S,2013
Chiò A[23]	2011	Arch Neurol	Same patient group as Borghero G,2014
Renton AE[24]	2011	Neuron	Same patient group as Majounie E,2012
Tai CP[25]	2011	Neurobiol Aging	Same patient group as Soong BW,2014
Boeve BF[26]	2012	Brain	Same patient group as DeJesus -Hernandez M,2011
Tsai CP[27]	2012	Neurobiol Aging	Same patient group as Soong BW,2014
van Blitterswijk M[28]	2012	PLoS One	Same patient group as van Blitterswijk M,2012
van Rheenen W[29]	2012	Neurology	Same patient group as van Blitterswijk M,2012
Zou ZY[30]	2012	Neurobiol Aging	Same patient group as Zou ZY,2016
Zou ZY[31]	2012	Eur J Neurol	Same patient group as Zou ZY,2016
Alavi A[32]	2013	Neurobiol Aging	Same patient group as Alavi A,2014
Tang L[33]	2013	Chin J Neurol (Chinese)	Same patient group as He J,2015
Zou ZY[34]	2013	Neurobiol Aging	Same patient group as Zou ZY,2016
Zou ZY[35]	2013	Neurobiol Aging	Same patient group as Zou ZY,2016
Kim YE[36]	2016	Neurobiol Aging	Same patient group as Kwon MJ,2012
Andersen PM[37]	2003	Amyotrophic Lateral Sclerosis	Unclear familial history
Van Deerlin VM[38]	2008	Lancet Neurol	Unclear familial history
Brettschneider J[39]	2012	Acta Neuropathol	Uncomplete familial history
Brown JA[40]	2012	Amyotroph Lateral Scler	Unclear familial history
Beck J[41]	2013	Am J Hum Genet	Unclear familial history
Dombroski BA[42]	2013	JAMA Neurol	Unclear familial history

Fratta P[43]	2015	Neurobiol Aging	Unclear familial history
Gitcho MA[44]	2008	Ann Neurol	Enrollment bias due to excluding other mutation carrier
Chiò A[45]	2009	Neurobiol Aging	Enrollment bias due to excluding other mutation carrier
Corrado L[46]	2009	Hum Mutat	Enrollment bias due to excluding other mutation carrier
Ticozzi N[47]	2009	Neurology	Enrollment bias due to excluding other mutation carrier
Vance C[48]	2009	Science	Enrollment bias due to excluding other mutation carrier
Blair IP[49]	2010	J Neurol Neurosurg Psychiatry	Enrollment bias due to excluding other mutation carrier
Chiò A[50]	2010	Arch Neurol	Enrollment bias due to excluding other mutation carrier
Hewitt C[51]	2010	Arch Neurol	Enrollment bias due to excluding other mutation carrier
Origone P[52]	2010	Amyotroph Lateral Scler	Enrollment bias due to excluding other mutation carrier
Rademakers R[53]	2010	Muscle Nerve	Enrollment bias due to excluding other mutation carrier
Suzuki N[54]	2010	J Hum Genet	Enrollment bias due to excluding other mutation carrier
Chiang HH,2012[55]	2012	J Hum Genet	Enrollment bias due to excluding other mutation carrier
Chiò A[56]	2012	Brain	Enrollment bias due to excluding other mutation carrier
Orrù S[57]	2012	Clin Genet	Enrollment bias due to excluding other mutation carrier
Sabatelli M[58]	2012	Neurobiol Aging	Enrollment bias due to excluding other mutation carrier
Solski JA[59]	2012	Amyotroph Lateral Scler	Enrollment bias due to excluding other mutation carrier
Sproviero W[60]	2012	Neurobiol Aging	Enrollment bias due to excluding other mutation carrier
Xi Z[61]	2012	Arch Neurol	Enrollment bias due to excluding other mutation carrier
Smith BN[62]	2013	Eur J Hum Genet	Enrollment bias due to excluding other mutation carrier
Al-Sarraj S[63]	2011	Acta Neuropathol	Enrollment bias due to only including ALS/MND and FTD
Hübers A[64]	2015	Neurobiol Aging	Enrollment bias due to only including early onset ALS
Yulug IG[65]	1995	Hum Mol Genet	Selected screening of SOD1 gene
Ceroni M[66]	1999	Neurology	Selected screening of SOD1 gene
Kuzma-Kozakiewicz M[67]	2013	Amyotroph Lateral Scler Frontotemporal Degener	Mutation rate calculation not possible

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