

Supplementary Material

Comprehensive Genetic and Mutation Analysis of Familial Dementia with Lewy Bodies Linked to 2q35-q36

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Supplementary Table 1

Gene	Regulatory region ¹	Exon	Intron	Nucleotide change (relative to NT_005403.16)	Rs number	Amino acid substitution
DNAJB2 (+)			IVS1-110G>C	g.70353826G>C	rs908197	-
			IVS2-14C>T	g.70354702C>T	rs2276638	-
			IVS6+81C>T	g.70357148C>T	rs2276639	-
	3' RR			g.70361274C>T	rs6436131	-
EPHA4 (-)	5' RR			g.72646582C>A	rs16825430	-
			IVS7+108G>A	g.72530641C>T	rs6718949	-
			IVS7-49C>T	g.72529843G>A	rs2288627	-
			IVS12-29A>G	g.72510773T>C	rs3213844	-
			IVS16-40T>C	g.72500318A>G	rs2303901	-
		EX17		g.72500245C>T	rs35860178	p.Thr960
	3' UTR	EX17		g.72499443T>G	rs10196918	-
	3' UTR	EX18		g.72494684A>G	-	-
	3' UTR	EX18		g.72494408G>C	rs13385757	-
	3' UTR	EX18		g.72494102G>C	rs3770207	-
	3' UTR	EX18		g.72494062C>T	rs3770208	-
	3' UTR	EX18		g.72493693C>T	rs17370971	-
	3' UTR	EX18		g.72493450G>C	rs13433079	-
	3' UTR	EX18		g.72493193T>A	-	-
	3' UTR	EX18		g.72493036C>T	-	-
	3' UTR	EX18		g.72492930C>T	rs957465	-
	3' UTR	EX18		g.72492920C>T	rs957466	-
	3' UTR	EX18		g.72492697T>C	rs3087584	-
	3' UTR	EX18		g.72492416A>T	rs9758	-
	3' UTR	EX18		g.72492375G>T	rs8508	-
3' UTR	EX18		g.72492181C>T	rs3177117	-	
OBSL1 (-) ²	5' RR			g.70646497C>T	rs35118453	-
	5' RR			g.70646389A>G	rs11893842	-
	5' RR			g.70646282A>C	-	-
		EX1		g.70644791T>C	rs1061399	p.Leu194
		EX1		g.70644450A>G	rs10180675	p.Leu307
			IVS1-67A>G	g.70642529T>C	rs12623922	-
			IVS2-18T>A	g.70642125A>T	rs2292360	-
		EX4		g.70641430G>A	-	p.Phe606
			IVS4-71T>C	g.70641335A>G	rs4674412	-
		EX5		g.70641218G>A	-	p.Tyr628
				g.70641047G>T	rs1043537	p.Ala685
			IVS6-32T>A	g.70637797A>T	rs2303543	-
			IVS7+81A>G	g.70637412T>C	rs11904583	-
		EX8		g.70636811G>A	rs17859671	p.Asp894
		EX 8		g.70636762G>T	rs17855740	p.Ala910
			IVS9+228G>C	g.70635648C>G	rs907142	-
			IVS9-356G>T	g.70633991C>A	rs2129641	-
			IVS9-268C>T	g.70633903G>A	rs1039900	-
		EX12		g.70632190A>G	rs2278201	p.Pro1187
			IVS12-60G>C	g.70631816C>G	rs2278200	-
			IVS13+36C>T	g.70631444G>A	-	-
			IVS13-34T>C	g.70630895A>G	rs13010015	-
			IVS13-11G>T	g.70630872C>A	rs1983211	-
		EX14		g.70630833C>G	rs1983210	p.Glu1365Asp
		EX14		g.70630751G>A	-	p.Leu1393
		EX15		g.70630406C>T	-	p.Arg1454Gln
		EX15		g.70630372A>G	rs10932816	p.Asp1465
	EX15		g.70630338C>T	-	p.Ala1477Thr	
	EX15A		g.70629969C>T	rs1056908	-	
	EX15A		g.70629588T>C	rs35372421	-	
	EX15A		g.70629353A>C	rs10932815	-	
	EX15A	3' RR (isoform C)	g.70629303G>A	-	-	

Supplementary Table 1, continued

Gene	Regulatory region ¹	Exon	Intron	Nucleotide change (relative to NT_005403.16)	Rs number	Amino acid substitution	
STK16		EX16		g.70628755T>C	rs10932814	p.Gln1578Arg	
		EX16		g.70628652T>C	rs10932813	p.Thr1612	
			IVS16-148G>C	g.70627972C>G	rs6754985	-	
		EX19		g.70626772G>T	-	p.Thr1737Lys	
		EX19		g.70626682C>T	-	p.Arg1767Gln	
			IVS19+30C>A	g.70626644G>T	-	-	
			IVS19-29G>A	g.70626383C>T	rs4672934	-	
			IVS19-4C>G	g.70626358G>C	rs34490902	-	
			IVS21-270T>C	g.70625250A>G	rs6717394	-	
			IVS21-188G>A	g.70625168C>T	rs6745782	-	
			IVS21-153C>T	g.70625133G>A	rs3731918	-	
			IVS21-82A>G	g.70625062T>C	rs1043582	-	
			IVS21-32_-33delTC	g.70625016_70625017delGA	rs3832112	-	
			EX8		g.70322658T>G	rs7601549	p.Leu293
			EX8		g.70322800G>A	rs8447	-
	3' UTR		g.70323144G>A	rs885854	-		
	3' RR						
TUBA1-4A(-)							
	5' RR		g.70328089delG	rs17552767	-		
TUBA4-4B(+)							
		EX4		g.70345767G>A	rs3731892	-(pseudogene)	
		EX4		g.70345787T>C	rs3731894	-(pseudogene)	
		EX4		g.70346252G>T	rs10166888	-(pseudogene)	
ABCB6(-)							
		EX1		g.70292695C>T	rs1109866	p.Leu39	
		IVS8+46C>G		g.70288212G>C	-	-	
		IVS9+50A>G		g.70287912T>C	rs2139823	-	
		IVS13-191G>A		g.70286821C>T	rs13027935	-	
APEG1(+)							
	5'RR			g.70508688G>T	rs907684	-	
	5'RR			g.70508957G>T	rs907683	-	
		IVS1+248G>A		g.70509787G>A	rs745027	-	
		IVS1+308G>A		g.70509811G>A	rs7576601	-	
		IVS1-889insTG		g.70517900_70517901insTG	-	-	
		IVS1+793A>C		g.70517997A>C	rs10932805	-	
		EX5		g.70525386G>A	rs10932806	p.Lys742	
		IVS5-42C>T		g.70525729C>T	-	-	
		EX13		g.70543365T>C	rs10167209	p.Pro1188	
		IVS13-73C>T		g.70545932C>T	-	-	
		IVS15+143G>A		g.70546611G>A	rs1039897	-	
		EX20		g.70551419G>A	-	p.Glu1522	
		EX30		g.70558166C>T	rs10755037	p.Pro2189Leu	
		IVS31+253C>T		g.70559867C>T	rs1810144	-	
		IVS31+303A>G		g.70559917A>G	rs10167337	-	
		EX32		g.70562428G>A	rs875098	p.Pro2613	
		IVS32+72A>G		g.70562519A>G	rs875097	-	
		IVS32-59G>C		g.70562576G>C	rs11892048	-	
		IVS33+53G>T		g.70562855G>T	-	-	
		EX34		g.70562947C>A	rs13026308	p.Pro2687Thr	
		EX36		g.70563523A>G	-	p.Arg2790Gly	
		EX36		g.70563780C>T	-	p.Phe2875	
		EX36		g.70563957G>A	-	p.Pro2934	
		IVS36+96G>C		g.70564172G>C	rs6742231	-	
		IVS36+103_104delG		g.70564180_70564181delG	-	-	
		EX38		g.70564944A>G	rs12464085	p.His3079Arg	
		IVS38-67G>C		g.70565864G>C	rs6747041	-	
		EX39		g.70565935G>T	rs12473286	p.Pro3130	
		IVS39+41C>T		g.70566047C>T	rs6747090	-	
		IVS39-27C>T		g.70566221C>T	rs907146	-	

Supplementary Table 1, continued

Gene	Regulatory region ¹	Exon	Intron	Nucleotide change (relative to NT_005403.16)	Rs number	Amino acid substitution
ATG9A (-)	3'UTR	EX41		g.70567010C>A	rs1078212	-
	3'UTR	EX41		g.70567613C>T	rs1050816	-
	5'RR			g.70304088T>G	-	-
	5'RR			g.70303966G>A	-	-
				g.70299101C>G	rs2276637	-
				g.70299009_70299010insG	-	-
IHH (-)		EX14	IVS6+107G>C	g.70295260A>G	rs2276633	p.Thr775
	3'UTR	EX15		g.70294354C>T	rs10799	-
	3'UTR	EX15		g.70294318G>A	rs3755048	-
	3'UTR	EX15		g.70293766C>T	rs6436126	-
	5'RR			g.70135583T>C	rs437512	-
	5'RR			g.70135636A>G	rs7592246	-
PTPRN (-)		EX3		g.70129828T>C	rs3731881	p.Pro251
		EX3		g.70129453A>G	rs394452	p.Thr376
	3'UTR	EX3		g.70129170C>G	rs3099	-
			IVS3-54G>A	g.70378023C>T	rs2271593	-
PAX3 (-)			IVS3-62G>A	g.70378032C>T	rs2271594	-
			IVS16-67G>A	g.70370722C>T	rs2292604	-
			IVS20+89A>G	g.70365518T>C	-	-
	3'UTR	EX23		g.70364060_70364061insGG	-	-
			IVS1-91A>C	g.73371440T>G	rs6754024	-
		EX2		g.73371305A>G	rs12623857	p.Gly43
DES (+)			IVS2+73T>C	g.73371040A>G	rs12694578	-
			IVS2+91G>T	g.73371022C>A	rs13405641	-
			IVS3+129A>C	g.73369534C>T	rs28546421	-
			IVS3-98C>A	g.73368535G>T	rs2303948	-
			IVS5+88G>C	g.73306125G>C	rs1965791	-
		EX6		g.73295371C>A	rs2234675	p.Thr315
	3'UTR	EX10		g.73275222G>T	-	-
	3'UTR	EX10		g.73275214A>G	-	-
	3'UTR	EX10		g.73274183G>A	-	-
	5'RR			g.70491929T>C	rs2854885	-
	5'RR			g.70492371C>T	rs2070927	-
		EX1		g.70492675A>G	rs1318299	p.Pro25
	EX1		g.70492693T>C	rs2017800	p.Ser31	
SLC23A3 (-)			IVS1+64T>G	g.70493242T>G	rs907677	-
			IVS1-37C>T	g.70494195C>T	rs12991025	-
		EX4		g.70494725C>T	rs1058261	p.Asp276
		EX5		g.70495082G>C	rs12920	p.Leu338
		EX6		g.70495558G>A	rs1058284	p.Ala368
	3'UTR	EX9		g.70500889T>C	rs11685408	-
			IVS3-5T>C	g.70243246A>G	rs3795986	-
			IVS8+55T>C	g.70238316A>G	rs6753739	-
	3'UTR	EX11		g.70235785G>T	rs6713887	-
	ANKZF1 (+)	5'UTR	EX1	IVS12+12C>A	g.70303966G>A	-
CHPF (-)				g.70309968C>A	-	-
	3'UTR	EX13		g.70310203C>T	rs2293079	-
	5'RR			g.70618115T>G	rs907145	-
	5'RR			g.70618054G>A	rs745303	-
			IVS1+211T>C	g.70617152A>G	rs11685912	-
		IVS2+99A>C	g.70615655T>G	rs13004994	-	
		IVS3+144G>C	g.70614939C>G	-	-	

Supplementary Table 1, continued

Gene	Regulatory region ¹	Exon	Intron	Nucleotide change (relative to NT_005403.16)	Rs number	Amino acid substitution
DNPEP (-)	3'RR	EX2		g.70616138T>C	rs3731912	p.Leu168
		EX4		g.70614737T>C	rs6436155	p.Gln371Arg
		EX4		g.70614646G>A	rs6436154	p.His401
		EX4		g.70614142G>A	rs1043832	p.His569
		EX4		g.70614064C>T	rs1043833	p.Leu595
		EX4		g.70613797A>G	rs1976616	p.Tyr684
				g.70613068T>C	rs907144	-
			IVS4-75A>G	g.70460624T>C	rs1040009	-
			IVS4-45A>G	g.70460594T>C	rs2271457	-
			IVS5-80_-81delC	g.70460316_70460317delG	rs11352428	-
			IVS5-6C>T	g.70460242G>A	rs2271456	-
			IVS7+31G>T	g.70459738C>A	rs878437	-
		EX8		g.70459563A>G	rs907679	p.Val248Ala
			IVS9+161T>C	g.70458343A>G	rs10804272	-
			IVS11-68G>T	g.70455789C>A	-	-
			IVS12+49A>G	g.70455634T>C	rs6436148	-
			IVS14+16G>C	g.70448977C>G	rs3731905	-
		IVS14+43C>T	g.70448950G>A	rs3731904	-	
CRYBA2 (-)	5'RR			g.70068160G>A	rs12694444	-
GLB1L (-)						
STK11IP (+)			IVS1-70G>A	g.70317839C>T	rs35328001	-
			IVS4+44G>A	g.70316861C>T	-	-
			IVS4+55A>T	g.70316850T>A	-	-
			IVS13+55G>A	g.70312577C>T	-	-
		EX1		g.70672056G>T	rs681747	p.Arg1Met
INHA (+)			IVS3+37G>T	g.70675615G>T	rs620698	-
			IVS10-82C>G	g.70680532C>G	rs633200	-
			IVS13-98A>T	g.70682086A>T	-	-
		EX15		g.70682771G>A	rs673951	p.Val563Ile
			IVS15+143C>T	g.70683069C>T	rs675291	-
		EX18		g.70685859C>T	rs627530	p.Ser752Phe
			IVS18-32T>C	g.70686061T>C	rs581547	-
			IVS19+228C>T	g.70686424C>T	rs630164	-
			IVS22+91C>A	g.70688471C>A	rs2305052	-
			IVS23+44T>C	g.70688728T>C	rs624773	-
			IVS24-73G>C	g.70690109G>C	rs665845	-
		5'RR		g.70646173A>T	-	-
		5'UTR	EX1	g.70646389A>G	rs11893842	-
	5'UTR	EX1	g.70646497T>C	rs35118453	-	
			g.70648745G>A	-	-	
		EX2	g.70649094C>T	rs12720063	p.Ala177	
		EX2	g.70649332G>A	rs12720062	p.Ala257Thr	
NHEJ1 (+)			IVS1+134G>A	g.70234724C>T	rs3731890	-
			IVS2-29A>T	g.70231851G>C	rs2030452	-
			IVS3+18G>A	g.70231592C>T	rs6436120	-
			IVS3+120T>C	g.70231490A>G	rs12694451	-
			IVS3-59C>G	g.70221992G>C	rs7585742	-
			g.70151354C>T	-	-	
	3'UTR	EX8	g.70150479G>A	rs897477	-	
	3'UTR	EX8	g.70149900C>T	rs359960	-	
ACCN4 (+)		EX1	g.70588976C>G	rs3731909	p.Ala165	
			IVS5+150T>C	g.70607227T>C	rs6731344	-

Supplementary Table 1, continued

Gene	Regulatory region ¹	Exon	Intron	Nucleotide change (relative to NT_005403.16)	Rs number	Amino acid substitution	
SLC4A3 (+)	3'RR	EX6	IVS5+164C>T	g.70607241C>T	rs6716083	-	
				g.70609367A>G	rs2276643	p.Thr505	
				g.70611148G>A	rs3770237	-	
				g.70611167G>A	rs2385538	-	
			g.70612040G>T	rs11689270	-		
			g.70612081C>G	rs6436153	p.Gln633Pro		
			g.70612087G>T	rs11689281	p.Leu635Arg		
			g.70612096T>C	rs11695248	p.Val638Ala		
			g.70613068T>C	rs907144	-		
			EX4	IVS3+94T>A	g.70702801T>A	-	-
		g.70703534A>C		rs597306	p.His157Pro		
		IVS5-180_-181delG		g.70704021_70704022delG	rs3214742	-	
		IVS6+93A>C		g.70704583A>C	rs612078	-	
		IVS7-32C>T		g.70706367C>T	-	-	
		IVS10+143G>A		g.70707737G>A	-	-	
		IVS10+171G>C		g.70707765G>C	-	-	
		IVS12+173G>A		g.70708914G>A	-	-	
		IVS12-101C>T		g.70709307C>T	-	-	
		IVS13-74C>G		g.70709737C>G	rs2305055	-	
		EX14		g.70709839A>G	rs589767	p.Glu694	
			g.70709842A>G	rs639214	p.Ala695		
	EX21		g.70710246G>C	rs35394392	-		
			g.70712548C>A	-	-		
			g.70714681T>C	rs684428	p.Leu1158		
			g.70714836C>T	-	-		
	3'UTR	EX23	IVS21+90C>T	g.70715938G>A	rs2305054	-	
ZFAND2B (+)			IVS1+39C>G	g.70281219C>G	rs6714820	-	
			IVS1+124_+125delG	g.70281303_70291304delG	-	-	
			IVS4+6G>T	g.70282182G>T	rs4674399	-	
			IVS4+92C>T	g.70282268C>T	rs4674367	-	
GMPPA (+)	5'RR	EX1		g.70570759G>A	rs2385540	-	
				g.70570850C>G	rs1567489	-	
				g.70570872A>T	-	-	
				g.70570901G>A	rs2010592	-	
				g.70570935C>G	rs16859986	-	
				g.70571005_70571006insA	-	-	
				g.delACGTCA	-	-	
				g.70571357G>A	-	-	
				g.70571698C>G	rs2010528	-	
				g.70571972C>T	rs12474050	-	
				g.70572314C>G	rs2029251	-	
				g.70572903A>G	rs2280039	-	
				g.70573332G>A	rs719337	-	
				g.70576368G>C	rs2385539	-	
RESP18 (-)	5'RR	EX1	IVS5-151G>C	g.70576368G>C	rs2385539	-	
				IVS10-103G>T	g.70580014G>T	-	-
				IVS12+37T>A	g.70580596T>A	rs10932808	-
				IVS1+32_+47del16	g.70407251_70407266del16	-	-
			IVS1+153C>G	g.70407145G>C	rs2385400	-	
			IVS1+211T>C	g.70407087A>G	rs2385401	-	
			IVS1-32G>C	g.70406907C>G	rs2385402	-	
			EX2		g.70406839C>T	rs2385403	p.Pro18
					g.70406803C>A	rs2385404	p.Glu30Asp
			EX2		g.70406736G>A	rs2385405	p.Glu11Lys
		g.70406622T>C		rs2048737	-		
	5'RR		IVS2+39A>G	g.70406022C>T	rs7562814	-	
			IVS2+639G>A	g.70406022C>T	rs7562814	-	
	5'RR		IVS2+678C>A	g.70405983G>T	rs1877867	-	

Supplementary Table 1, continued

Gene	Regulatory region ¹	Exon	Intron	Nucleotide change (relative to NT_005403.16)	Rs number	Amino acid substitution		
CCDC108 (-)	5'RR		IVS2-660T>C	g.70405672A>G	rs1567932	-		
			IVS3+47G>C	g.70404861C>G	rs6707009	-		
			IVS1+144G>T	g.70115481C>A	rs1167440	-		
			IVS2-15A>G	g.70113202T>C	rs4672910	-		
		EX3 EX4		g.70113138C>T g.70112673T>G	rs4674354 rs6736922	p.Val16 p.Met66Leu		
C2orf24 (-)	5'RR 5'RR 5'RR 5'RR 5'RR		IVS24-77G>A IVS27-139C>T	g.70085210C>T g.70083736G>A	- -	- -		
				g.70252326_70252327insTG	-	-		
				g.70252322G>A	-	-		
				g.70252073G>A	rs10170244	-		
				g.70252042A>C	rs10193576	-		
C2orf17 (+)	5'UTR		IVS1+27G>A IVS1+49G>T IVS1-171_-170delAT IVS1-38C>T	g.70250870C>T g.70250848C>A g.70250639_70250640delTA g.70250506G>A	- - rs59013810 rs6436123	- - - -		
		EX3		g.70249745A>G	rs1127101	p.Tyr81		
		EX8		g.70247171A>G	rs1043160	p.Ile262Thr		
		EX8		g.70247081A>G	rs1127102	p.Leu292Pro		
		EX8		g.70246808A>G	rs4674361	p.Leu383Pro		
		EX8		g.70246512C>T	rs11236	-		
		ENSG188760(+)	5'RR 5'RR 5'UTR 5'UTR		IVS2-180A>C IVS7-13G>C	g.70252090G>A g.70253690A>C g.70255764G>C	rs10170244 - rs2272017	- - -
				EX9		g.70256390C>A	rs3731900	p.Pro419Gln
				EX9		g.70256808C>T	rs3731901	-
				EX9		g.70257565C>T	rs7017	-
				EX9		g.70257808G>A	-	-
		miR-153-1 (-)	5'RR 5'RR 5'UTR 5'UTR		IVS3+84C>T	g.70617151A>G g.70617333G>T g.70618053G>A g.70618114T>G g.70619116C>T	rs11685912 rs10932810 rs745303 rs907145 rs10932811	- - - - -
EX4				g.70621671T>C	rs8179770	p.Phe65		
EX5				g.70623431C>T	rs6436156	p.Asp295		
EX5				g.70623434T>C	rs13023533	p.Pro296		
				g.70368703G>C	-	-		
				g.72070557A>G	-	-		