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Supplementary Data

Contribution of TARDBP to Alzheimer's Disease Genetic Etiology

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Supplementary Table 1

TARDBP sequence variants in Belgian AD patients

Location	gDNA[1]	cDNA[2]	protein[3]	rs-nr	MAF[4](%)
5' upstream region	g.6825delC	c.-134-145delC	–	–	0.2
5' upstream region	g.6870T>C	c.-134-100T>C	–	rs968545	4.8
EX 1	g.7024delG	c.-80delG	–	–	0.1
EX 1	g.7033C>G	c.-71C>G	–	–	0.1
IVS 1	g.7176C>T	c.-13+85C>T	–	rs4133584	17.1
EX 2	g.8273T>C	c.198T>C	p.Ala66	rs61730366	0.4
IVS 2	g.11177G>A	c.239-15G>A	–	–	0.1
EX 3	g.11222C>T	c.269C>T	p.Ala90Val	–	0.2[5]
IVS 3	g.12921A>G	c.403-160A>G	–	–	0.1
IVS 4	g.13333C>A	c.543+112C>A	–	–	0.1
IVS 4	g.13368A>G	c.543+147A>G	–	rs2273348	17.4
EX 5	g.14875C>T	c.642C>T	p.Tyr214	–	0.1
IVS 5	g.15016_15017insG	c.714+69_70insG	–	rs70977536	15.6
IVS 5	g.15083G>A	c.714+136G>A	–	–	0.1
IVS 5	g.16346delG	c.715-126delG	–	rs3835416	17.4[6]
IVS 5	g.16398_16399insT	c.715-74_-73insT	–	–	0.8[6]
EX 6	g.16732C>T	c.975C>T	p.Ala325	–	0.1[6]
EX 6	g.16879T>C	c.1122T>C	p.Tyr374	–	0.1[6]

[1]gDNA numbering relative to GenBank accession number AL109811.40 and starting at nucleotide 1.
[2]cDNA numbering according to the largest *TARDBP* transcript with GenBank Accession Number NM_007375.3 and starting at the translation initiation codon.
[3]Protein numbering according to the largest*TARDBP* isoform with GenPept Accession Number NP_031401.1.
[4]MAF: minor allele frequency, calculated in the group of 485 AD patients. Exceptions: [5]pAla90Val where MAF is calculated in a total of 739 patients and [6]calcluated in 675 patients.

Supplementary Table 2

Allele sharing in the *TARDBP* locus in p.Ala90Val carriers

marker	Physical location (Mb)[1]	Frequency shared allele[2]	DR146B[3]	DR520.1		DR538.1	
D1S2663	7.18	–	**176**	172	174	174	182
D1S2694	7.26	–	**412**	421	423	423	425
D1S2736	10.54	–	**129**	123	123	**129**	**129**
STRtel7	**10.78**	**0.78**	**131**	**131**	133	**131**	**131**
STRtel5	**10.89**	**0.50**	**453**	**453**	455	**453**	**453**
D1S1635	**10.91**	**0.17**	**405**	393	**405**	393	**405**
TARDBP **p.Ala90Val**	**11.00**	–	**T**	C	**T**	C	**T**
D1S2667	**11.41**	**0.33**	**128**	**128**	**128**	124	**128**
STRcen4	**11.48**	**0.39**	**391**	389	**391**	381	**391**
D1S2470	**11.84**	**0.02**	**136**	**136**	146	**136**	146
D1S434	12.25	–	**250**	248	248	248	248
D1S1597	13.66	–	**348**	332	**348**	332	353
D1S402	14.04	–	**428**	404	436	404	416
D1S407	14.73	–	**445**	**445**	453	449	453
D1S2728	15.03	–	**247**	**247**	251	249	251
D1S2672	15.15	–	**151**	147	**151**	147	**151**
D1S3669	17.66	–	**182**	**182**	202	186	202

Shared alleles are indicated in bold.
[1]Physical location of STR markers relative to NCBI genome build 36.
[2]Frequencies of the shared alleles were calculated in at least 90 control individuals.
[3]p.Ala90Val haplotype determined by segregation analysis of relatives of DR146B.