Table e-1. Characteristics of *GBA* mutation carriers

Subject	Disease group	*GBA* mutation	*APOE*[a]	Race	Age at onset	Age at death	Sex	ADNCs		LBDNCs							LBDNC stage[b]
								Braak stage	CERAD score	MD	SN	AG	HP	PHG	CG	FC	
Pathogenic mutations																	
1	Control	H255Q	2.3	W	--	74	M	1	0	–	–	–	–	–	–	–	None
2	Control	IVS2+1	3.3	W	--	89	F	3	A	–	–	–	–	–	–	–	None
3	Control	N370S	3.3	W	--	71	M	0	0	NA	–	NA	NA	–	–	–	None
4	AD	N370S	4.4	W	79	86	F	5	C	–	–	–	–	–	–	–	None
5	AD	R496H	3.4	W	89	95	F	6	C	–	–	–	–	–	–	–	None
6	LBD-AD	Rec1[c]	3.4	W	67	73	M	6	C	+	+	+	+	+	+	+	Neocortical
7	LBD-AD	N370S	3.3	W	74	78	M	5	C	+	+	+	+	+	+	+	Neocortical
8	LBD-AD	N370S	3.3	W	84	90	M	5	C	+	+	+	+	+	+	+	Neocortical
9	LBD-AD	N370S	3.4	W	66	72	F	4	B	+	+	+	+	+	+	+	Neocortical
10	LBD-AD	N370S	3.3	W	79	87	M	6	C	+	+	+	+	+	+	+	Neocortical
11	LBD-AD	N370S	3.4	W	68	76	M	4	C	+	+	+	+	+	+	+	Neocortical
12	LBD-AD	Rec1[c]	3.3	W	85	87	M	4	B	NA	+	NA	NA	+	+	–	Limbic
13	LBD-AD	Rec1[c]	3.3	W	87	90	F	4	B	NA	+	NA	NA	+	+	+	Neocortical
14	pDLB	D140H	2.4	W	85	89	M	1	A	NA	+	NA	NA	+	–	–	Limbic
15	pDLB	L444P	3.4	W	52	57	M	1	A	+	+	+	NA	NA	+	+	Neocortical
16	pDLB	N370S	2.4	W	63	79	M	1	0	+	+	+	+	+	+	+	Neocortical
17	pDLB	N370S	3.3	W	57	65	M	2	0	+	+	+	+	+	+	+	Neocortical
18	pDLB	N370S	4.4	W	69	73	M	2	A	+	+	+	+	+	+	+	Neocortical
19	pDLB	N370S	4.4	W	54	79	M	2	A	+	+	+	+	+	+	+	Neocortical

Mutations of Unknown Significance																	
20	Control	R262H	3.4	W	--	81	F	2	0	NA	–	NA	NA	–	–	–	None
21	AD	H422Y	3.3	W	76	86	F	6	C	–	–	–	–	NA	NA	–	None
22	LBD-AD	R39C	3.3	W	88	95	M	4	C	+	+	+	+	+	+	+	Neocortical
23	LBD-AD	R262H	3.3	W	70	82	F	6	C	+	+	+	+	+	+	+	Neocortical
24	LBD-AD	E388K	3.3	W	75	81	F	6	C	+	+	+	+	+	NA	+	Neocortical
25	pDLB	E388K	3.3	W	72	80	M	2	A	+	+	+	+	+	+	+	Neocortical

AD, dementia with high-level Alzheimer's disease neuropathologic changes without Lewy body disease neuropathologic changes; ADNCs, Alzheimer's disease neuropathologic changes; AG, amygdala; CERAD, Consortium to Establish a Registry for Alzheimer's Disease; CG, cingulate gyrus; FC, frontal cortex; HP, hippocampus; LBD-AD, dementia with Lewy body disease neuropathologic changes and high-level Alzheimer's disease neuropathologic changes; LBDNCs, Lewy body disease neuropathologic changes; MD, medulla; NA, not available; pDLB, pure dementia with Lewy bodies (dementia with Lewy body disease neuropathologic changes and no or low-level Alzheimer's disease neuropathologic changes); PHG, parahippocampal gyrus; SN, substantia nigra; W, white.

[a]*APOE* genotype
[b]Based on Leverenz et al. (2008)
[c]Recombinant allele containing L444P, A456P, and V460V