Mutational analysis in familial Alzheimer's disease of Han Chinese in Taiwan with a predominant mutation *PSEN1* p.Met146Ile

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Supplementary table S1. Mutational and clinical information of the index patients

No.	Gene	Exon	Nucleotide	Amino acid	Method	AAO (year)	mGS†	Sex	Initial symptoms	Additional neurological feature	Age of MRI (year)	MTA	PA	WMH
1	*PSEN1*	5	c.350C>T	p.Pro117Leu	SSCP	36	3	M	Memory	Myoclonus Emotional liability	38	0	0	0
2	*PSEN1*	5	c.392A>G	p.His131Arg	NGS	49	1	M	Memory Topo‡	-	61	2	1	1
3	*PSEN1*	5	c.471G>T	p.Arg157Ser	NGS	51	4	M	Memory	-	59	1	2	0
4	*PSEN1*	5	c.438G>A	p.Met146Ile	SSCP	45	1	F	Memory	-	-	-	-	-
5	*PSEN1*	5	c.438G>A	p.Met146Ile	SSCP	45	1	F	Topo	-	45	-	0	1
6	*PSEN1*	5	c.438G>A	p.Met146Ile	SSCP	40	1	F	Memory	Seizure EPS§	47	-	3	0
7	*PSEN1*	5	c.438G>A	p.Met146Ile	SSCP	48	1	M	Memory	-	48	1	1	0
8	*PSEN1*	5	c.438G>A	p.Met146Ile	SSCP	46	1	M	Memory	-	-	-	-	-
9	*PSEN1*	5	c.438G>A	p.Met146Ile	SSCP	40	1	F	Memory Topo	Myoclonus Emotional liability	46	1	0	0
10	*PSEN1*	5	c.438G>A	p.Met146Ile	SSCP	43	2	M	Language Dysexecutive	-	46	1	2	0
11	*PSEN1*	5	c.438G>A	p.Met146Ile	SSCP	46	2	M	Memory	Myoclonus	46	0	0	0
12	*PSEN1*	5	c.438G>A	p.Met146Ile	SSCP	44	1	M	Memory	Myoclonus Seizure Choreoathetosis	55	2	1	0

									Auditory hallucination					
13	*PSEN1*	7	c.617G>A	p.Gly206Asp	SSCP	33	3	F	Memory	Seizure	-	-	-	-
14	*PSEN1*	7	c.626G>A	p.Gly209Glu	SSCP	60	2	M	Memory	Apraxia	64	2	1	0
15	*PSEN1*	8	c.838G>A	p.Glu280Lys	NGS	55	4	F	Memory	-	-	-	-	-
16	*PSEN1*	8	c.856C>G	p.Leu286Val	SSCP	43	1	M	Memory	-	-	-	-	-
17	*APP*	16	c.2066G>C	p.Asp678His	SSCP	50	4	F	Memory	-	-	-	-	-

mGS, modified Goldman score; AAO, age at onset; MTA, medial temporal atrophy; PA, posterior atrophy; WMH, white matter hyperintensity; SSCP, single strand-conformational polymorphism with subsequent Sanger sequencing; NGS, next generation sequencing

† A modified Goldman score of 1 is defined by the presence of at least three affected people in two generations, with one person being a first-degree relative of the other two; a score of 2 is familial aggregation of three or more family members with dementia not meeting the criteria for a score of 1; a score of 3 is one other affected family member with dementia (modified to give a score of 3 only if there is a history of young-onset dementia within the family, i.e., AAO less than 65 years; with a score of 3.5 if AAO is above 65); and a score of 4 is no or an unknown family history.

‡Topo, topographic disorientation

§EPS, extrapyramidal symptoms