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Supplementary Data

PSEN1 Mutation Carriers Present Lower Cerebrospinal Fluid Amyoid-β_{42} Levels than Sporadic Early-Onset Alzheimer's Disease Patients but no Differences in Neuronal Injury Biomarkers

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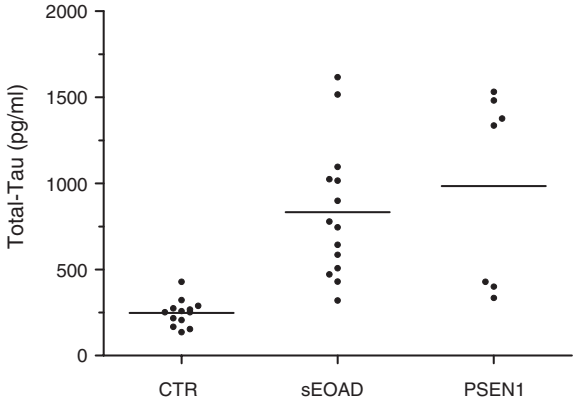

Supplementary Figure 1. Distribution of CSF t-tau values in the different study groups. PSEN1 = *PSEN1* mutation carriers; sEOAD = sporadic early-onset AD; CTR = controls.

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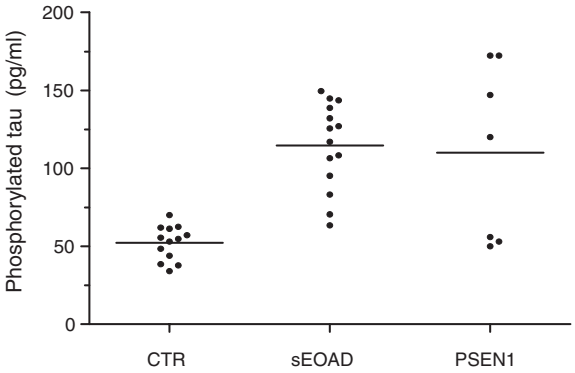

Supplementary Figure 2. Distribution of CSF p-tau values in the different study groups. PSEN1 = *PSEN1* mutation carriers; sEOAD = sporadic early-onset AD; CTR = controls.

Supplementary Table 1
Mean group values and post-hoc comparisons between groups. Estimated intracranial volume (EIV) and subcortical values are volume estimations, whereas the cortical region of interest (ROI) values are expressing cortical thickness measures

	CTR	sEOAD	PSEN1	CTR versus sEOAD	CTR versus PSEN1	sEOAD versus PSEN1
EIV	1441426 (160518)	1408693 (180461)	1432314 (124750)	$p = 0.868$	$p = 0.992$	$p = 0.320$
Left superior frontal	2.762 (0.133)	2.620 (0.111)	2.587 (0.100)	$p = 0.013$*	$p = 0.008$*	$p = 0.820$
Left thalamus	6788 (830)	6311 (801)	6217 (597)	$p = 0.279$	$p = 0.265$	$p = 0.963$
Left hippocampus	4177 (598)	3299 (574)	3691 (562)	$p = 0.001$*	$p = 0.185$	$p = 0.327$
Right thalamus	7083 (865)	6307 (647)	6403 (606)	$p = 0.030$*	$p = 0.128$	$p = 0.958$
Right hippocampus	4280 (575)	3331 (636)	3723 (449)	$p = 0.001$*	$p = 0.109$	$p = 0.320$

sEOAD = sporadic early-onset AD; PSEN1 = carriers of *PSEN 1* mutations; CTR = controls. *$p < 0.05$.

Supplementary Table 2
Voxel-based morphometry (VBM) results for the controls versus AD group contrasts. Coordinates are shown according to the Montreal Neurological Institute (MNI) atlas. Only clusters with at least 30 voxels are shown

Contrast	Voxels	*t*-value	X (MNI)	Y (MNI)	Z (MNI)	Brain region
CTR > sEOAD	37166	5.18	−4	−56	24	Precuneus/PCC
	390	4.02	8	−68	−28	Cerebellum
CTR > PSEN1	36842	6.20	−54	−30	−14	MTC
	150	4.05	−36	6	32	L MFG
	148	3.23	−28	4	44	L MFG
	143	3.82	−14	20	36	L PaC
	41	3.31	−16	48	26	L Frontal Pole
sEOAD > PSEN1	388	4.58	70	−12	4	R STG
	369	2.33	36	−4	−30	R Fusiform Gyrus
	335	3.12	10	−22	16	Right Thalamus
	262	2.44	−32	−26	10	L Insular Cortex
	260	3.38	10	36	36	R PaC
	208	2.50	−4	20	20	L ACC
	184	2.75	14	−64	38	R Precuneus
	181	2.68	56	−72	−16	R LOC
	80	2.22	−10	−58	42	L Precuneus
	51	1.79	56	−42	−30	R ITG

SFG = superior frontal gyrus; MFG = middle frontal gyrus; PaC = paracingulate cortex; AAC = anterior cingulate cortex; STG = superior temporal gyrus; ITG = inferior temporal gyrus; LOC = lateral occipital cortex. Controls > sEOAD and Controls > PSEN1 contrasts are corrected by multiple comparisons and thresholded at $p < 0.05$. sEOAD > PSEN1 is a $p < 0.001$ uncorrected contrast.

Supplementary Table 3
Areas with significant values in cortical thickness comparisons (sEOAD versus CTR, PSEN1 versus CTR, CTh/t-tau correlation) after FWE corrections for multiple comparisons using Monte Carlo simulations (10000 permutations). Cortical areas corresponding to each cluster were assigned using the parcellations from an available atlas [12]

Comparison	Size (mm^2)	Max p value	Max-p MNI coordinates	Cluster-wise probability	Cluster-wise interval	Atlas label
Total tau – CTh correlation	3083.23	$10^{(-2.83)}$	(−27.47, 49.94, 43.88)	0.0003	(0.0001–0.0050)	Superior parietal
	1497.01	$10^{(-2.47)}$	(−27.17, −4.89, 44.25)	0.0429	(0.0403–0.04550)	Caudal middle frontal
SEOAD < CTR Left Hemisphere CTh	30936.43		(−27.47, 50.39, 37.89)	0.0001	(0.0000–0.0002)	Inferior parietal
	2693.51		(−35.45, 1.30, 31.30)	0.0008	(0.0005–0.0012)	Caudal middle frontal
sEOAD < CTR Right Hemisphere CTh	29985.29		(42.9293 −49.1, -12.25)	0.0001	(0.0000–0.0002)	Inferior temporal
	2443.39		(20.30, 25.46, 46.29)	0.0010	(0.0006–0.0014)	Superior frontal
PSEN1 < CTR Left Hemisphere CTh	35710.22		(−16.36, −71.72, 60.64)	0.0001	(0.0000–0.0002)	Superior parietal
PSEN1 < CTR Right Hemisphere CTh	33937.50		(44.14, −38.92, 2.29)	0.0001	(0.0000–0.0002)	Bank of superior temporal sulcus