Identification of a Late Onset Alzheimer’s Disease Candidate Risk Variant at 9q21.33 in Polish Patients

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Supplementary Figure 1. T-ARMS PCR results obtained for detected genotypes of rs429358 and rs7412 variants of the APOE gene. 514 bp: the amplicon generated by the outer primers present in all reactions; 444 bp: rs429358 C (130Arg), 307 bp: rs7412 C (176Arg), 253 bp: rs429358 A (130Cys), 115 bp: rs429358 A (130Cys). The patient DNA samples are shown against a GeneRuler™ DNA Ladder Mix, a 100–10,000 bp DNA fragment size marker (Fermentas; Thermo Fisher Scientific Inc., USA).

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Supplementary Figure 2. Statistical power of the GWAS for alleles found at different frequencies (p0) in the general population. Results for the cohorts including 141 (MCI+LOAD) and 94 (LOAD) case subjects are plotted in A and B, respectively.

Supplementary Figure 3. Principle component analysis of the pooled DNA microarray experiments based on unfiltered relative allele signal (RAS) intensities. Pool 1, mild cognitive impairment; Pools 2 and 3, LOAD; Pools 4–6, control. Each of the six pooled-DNA experiments was run in three technical repeats (A, B, C).

Supplementary Figure 4. Histograms of the relative allele signal (RAS) for six 47-patient pools: Pool 1, MCI; Pool 2 and 3, LOAD; Pool 4–6, control. Blue color denotes RAS histograms after raw signal filtration for each pooled DNA sample, while the red color represents the distribution of RAS values calculated using the signal intensities, which were filtered out from the raw datasets.
Supplementary Figure 5. Quantile-quantile plots of the χ² p-value distribution for the studies including 141 MCI+LOAD (A), and 94 LOAD (B) cases.
Supplementary Figure 6. Manhattan plots indicating association significance and chromosomal positions of the SNPs selected in pooled-DNA GWAS. MCI+LOAD versus N (A); LOAD versus N (B).