

Supplementary Material

Polygenic Risk Scores in Alzheimer’s Disease Genetics: Methodology, Applications, Inclusion, and Diversity

Supplementary Table 1. Alzheimer’s Disease PRS Results. Summary of Alzheimer’s Disease PRS papers mentioned in this review, with respective phenotypes, sample sizes, and association results.

| Author, Year | Training Pop. | Training Pheno | Testing Pop. | Testing Pheno | Sample Size (Case/Control or Total N) | Results | | |
|-------------------|---------------|----------------|---------------|--|---------------------------------------|------------------|-------------|----------|
| Marden, 2016 [1] | NHW | AD | NHW | Memory Decline | 7172 | p < 0.0001* | | |
| | NHB | | NHB | | 1081 | p = 0.080 | | |
| Mormino, 2016 [2] | NHW | AD | NHW | Baseline Memory | 166/2353 | p = 0.002* | | |
| | | | | Cognitive Decline (Memory) | | p = 0.0005* | | |
| | | | | Cognitive Decline (Executive Function) | | p = 0.01* | | |
| | | | | Baseline Hippocampal Function | | p = 0.002* | | |
| | | | | Clinical Progression (MCI/AD) | | p < 0.00001* | | |
| | | | | Abeta PET | | p = 0.03* | | |
| | | | | Longitudinal Hippocampal Function | | p = 0.06 | | |
| | | | | Baseline Executive Function | | p = 0.32 | | |
| | | | | CSF Abeta | | p = 0.11 | | |
| Desikan, 2017 [3] | NHW | AD | NHW | AAO | 6409/9386 | p = 1.1e-26* | | |
| | | | | Longitudinal Progression (AD) | | p = 1.5e-10* | | |
| | | | | Neuropathology (Braak Stage) | | p = 3.9e-6* | | |
| | | | | Neuropathology (CERAD Score) | | p = 6.8e-6* | | |
| | | | | Entorhinal Cortex Volume Loss | | p = 6.3e-6* | | |
| | | | | Hippocampal Volume Loss | | p = 7.9e-5* | | |
| Tosto, 2017 [4] | NHW | LOAD | NHW | LOAD AAO | 2128/4792 | OR = 1.29* | | |
| | Caribbean His | | Caribbean His | LOAD AAO | 2155/3324 | OR = 1.73* | | |
| Tan, 2018 [5] | NHW | AD | NHW | Amyloid load | 599/347 | p = 5.5e-5* | | |
| | | | | Neurofibrillary Tangles | | p = 6.53e-4* | | |
| Felsky, 2018 [6] | NHW | AD | NHW | Global Cognition | 25580/48466 | p = 2e-5* | | |
| | | | | Microglial Densities | | p = 0.51 | | |
| | | | | RA | | Global Cognition | 29880/73758 | p = 0.93 |
| | | | | Microglial Densities | | p = 1.6e-4* | | |

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|---------------|-----|-----------------|-----|--|--------------|---------------|
| | | MS | | Global Cognition | 47351/68284 | p = 0.45 |
| | | | | Microglial Densities | | p = 0.12 |
| | | PD | | Global Cognition | 13708/95282 | p = 0.71 |
| | | | | Microglial Densities | | p = 0.11 |
| | | CAD | | Global Cognition | 60801/123504 | p = 0.54 |
| | | | | Microglial Densities | | p = 0.27 |
| | | Schizophrenia | | Global Cognition | 36989/113075 | p = 0.42 |
| | | | | Microglial Densities | | p = 0.42 |
| | | Telomere Length | | Global Cognition | 48423 | p = 0.49 |
| | | | | Microglial Densities | | p = 0.27 |
| Tan, 2019 [7] | NHW | AD | NHW | Amyloid PET | 6409/9386 | FDR < 0.05* |
| | | | | Longitudinal Regional Cortical Volume Change | | FDR < 0.05* |
| | | | | Postmortem Amyloid-beta | | p = 7.29e-10* |
| | | | | Rate of Memory Decline | | p = 3.65e-3* |
| | | | | Rate of CDR-SB Decline | | p = 2.44e-10* |
| | | | | Rate of Executive Function Decline | | p = 2.94e-2 |

Asterisk (*) indicates a significant association. NHW, Non-Hispanic White; NHB, Non-Hispanic Black; AAO, age at onset; RA, rheumatoid arthritis; MS, multiple sclerosis; PD, Parkinson's disease; CAD, coronary artery disease.

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