



EMIF Deliverable 3.8: 4 Epigenomic profiles associated with abnormal AD CSF biomarkers

Executive summary

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The goal of this task was to identify differentially methylated regions (DMRs) associated with abnormal CSF biomarker levels generated by our project partners by performing an epigenome-wide association study (EWAS) using DNA methylation profiles as predictors.

To study the role of differential DNA methylation we used the “Infinium MethylationEPIC” array (Illumina, Inc.), which covers ~850K methylation sites across the genome. DNA methylation profiling was performed at UzL, all data processing, QC, and statistical analyses were performed at the University of Exeter (UNEXE).

The EWASs performed for the EMIF-AD study have identified several novel genes associated with diagnosis and peripheral and clinical measures, as well as some genes previously implicated in dementia (e.g. QKI and HLF gene expression in AD brain samples (Farnsworth et al, 2016; Silva et al, 2012)). These are currently subject to independent validation analyses. Current dissemination activities include preparing the abovementioned genomics results for peer-reviewed publication. Subsequent analyses will focus on generating and disseminating multivariate results (e.g. by integrating genomic and epigenomic data) in these same individuals studied in the different modalities in WP3.

Contacts

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