



Genomics for Social Scientists

Epigenetics

NATIONAL INSTITUTE ON AGING

Epigenome-wide association studies:

- DNA methylation signatures of chronic low-grade inflammation are associated with complex diseases.
<https://genomebiology.biomedcentral.com/articles/10.1186/s13059-016-1119-5>
- An epigenome-wide association study meta-analysis of educational attainment.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6372242/>
- Epigenome-wide association of myocardial infarction with DNA methylation sites at loci related to cardiovascular disease.
<https://clinicalepigeneticsjournal.biomedcentral.com/articles/10.1186/s13148-017-0353-3>
- Epigenome-wide association study of body mass index, and the adverse outcomes of adiposity.
<https://www.nature.com/articles/nature20784>
- Epigenome-wide association study for perceived discrimination among sub-Saharan African migrants in Europe - the RODAM study.
<https://www.nature.com/articles/s41598-020-61649-0>
- DNA methylation analysis identifies loci for blood pressure regulation.
<https://www.sciencedirect.com/science/article/pii/S0002929717304202?via%3Dihub>
- DNA methylation mediates the association between individual and neighborhood social disadvantage and cardiovascular risk factors.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC9162507/>

Other epigenetic application papers:

- DNA Methylation Mediates the Association Between Individual and Neighborhood Social Disadvantage and Cardiovascular Risk Factors
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC9162507/>

Epigenetic methods papers:

- Polymethylation scores for prenatal maternal smoke exposure persist until age 15 and are detected in saliva in the Fragile Families and Child Wellbeing cohort. This is an example of creating and applying scores from an existing EWAS.
<https://www.tandfonline.com/doi/abs/10.1080/15592294.2022.2112815>
- Methodological challenges in constructing DNA methylation risk scores.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6961658/>
- Comparison of Beta-value and M-value methods for quantifying methylation levels by microarray analysis.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3012676/>

- Controlling bias and inflation in epigenome- and transcriptome-wide association studies using the empirical null distribution.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5273857/>
- Bump hunting to identify differentially methylated regions in epigenetic epidemiology studies.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3304533/>
- Comb-p: software for combining, analyzing, grouping and correcting spatially correlated P-values.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3496335/>
- De novo identification of differentially methylated regions in the human genome.
<https://epigeneticsandchromatin.biomedcentral.com/articles/10.1186/1756-8935-8-6>
- “Gap hunting” to characterize clustered probe signals in Illumina methylation array data.
<https://epigeneticsandchromatin.biomedcentral.com/articles/10.1186/s13072-016-0107-z>
- Accounting for population stratification in DNA methylation studies.
<https://pubmed.ncbi.nlm.nih.gov/24478250/>
- Differential methylation between ethnic sub-groups reflects the effect of genetic ancestry and environmental exposures.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5207770/>
- Genome-wide methylation data mirror ancestry information.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5267476/>
- Accurate ethnicity prediction from placental DNA methylation data.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6688210/>
- SeSAME: reducing artifactual detection of DNA methylation by Infinium BeadChips in genomic deletions.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6237738/>

Epigenetic clock methods papers:

- A computational solution for bolstering reliability of epigenetic clocks: Implications for clinical trials and longitudinal tracking
<https://www.nature.com/articles/s43587-022-00248-2>
- Underlying features of epigenetic aging clocks in vivo and in vitro
<https://pubmed.ncbi.nlm.nih.gov/32930491/>
- DNA methylation-based biomarkers and the epigenetic clock theory of ageing
<https://www.nature.com/articles/s41576-018-0004-3>
- DNA methylation age of human tissues and cell types
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4015143/>
- Genome-wide methylation profiles reveal quantitative views of human aging rates
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3780611/>
- An epigenetic biomarker of aging for lifespan and healthspan
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5940111/>
- DNA methylation GrimAge strongly predicts lifespan and healthspan
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6366976/>
- Quantification of the pace of biological aging in humans through a blood test, the DunedinPoAm DNA methylation algorithm

- <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7282814/>
- DunedinPACE, a DNA methylation biomarker of the pace of aging
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC8853656/>
- DNA Methylation Clocks in Aging: Categories, Causes, and Consequences
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6520108/>

Epigenetic clock application papers:

- Associations of Age, Sex, Race/Ethnicity, and Education With 13 Epigenetic Clocks in a Nationally Representative U.S. Sample: The Health and Retirement Study:
<https://pubmed.ncbi.nlm.nih.gov/33453106/>
- A systematic review of biological, social and environmental factors associated with epigenetic clock acceleration:
<https://pubmed.ncbi.nlm.nih.gov/33930583/>
- Socioeconomic position, lifestyle habits and biomarkers of epigenetic aging: a multi-cohort analysis.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6503871/>
- Trends in DNA methylation with age replicate across diverse human populations.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5500158/>
- An epigenetic clock analysis of race/ethnicity, sex, and coronary heart disease.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4980791/>
- The role of epigenetic aging in education and racial/ethnic mortality disparities among older U.S. Women
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6555423/>
- The Socioeconomic Gradient in Epigenetic Ageing Clocks: Evidence from the Multi-Ethnic Study of Atherosclerosis and the Health and Retirement Study
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC9235889/>

Combining epigenetics and genetics:

- Improving phenotypic prediction by combining genetic and epigenetic associations.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4572498/>
- Mapping eQTL by leveraging multiple tissues and DNA methylation.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5648503/>
- Integrative analysis of multi-omics data for discovery and functional studies of complex human diseases.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5742494/>
- Two-step epigenetic Mendelian randomization: a strategy for establishing the causal role of epigenetic processes in pathways to disease.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3304531/>
- Genome-wide analyses of sparse mediation effects under composite null hypotheses.
<https://doi.org/10.1214/18-AOAS1181>
- Large-Scale Hypothesis Testing for Causal Mediation Effects with Applications in Genome-wide Epigenetic Studies.
<https://doi.org/10.1080/01621459.2021.1914634>

Online resources

- [Gene Expression Omnibus](#): a public functional genomics data repository supporting MIAME-compliant data submissions. Array- and sequence-based data are accepted. Tools are provided to help users query and download experiments and curated gene expression profiles.
- [Bioconductor](#): provides tools for the analysis and comprehension of high-throughput genomic data
- [Github](#): code hosting platform for version control and collaboration
- mQTL databases and similar resources for site look-ups:
 - *GoDMC - [Blood] <http://mqtl.db.godmc.org.uk/>
 - mQTLdb - [Blood] <http://www.mqtl.db.org/>
 - iMETHYL [Blood] <http://imethyl.iwate-megabank.org/>
 - CRIC: Human whole blood mQTL Atlas – [Diabetic kidney disease] <http://www.susztaklab.com/mqtl/index.php>
 - [Placenta] - <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5654961/>
 - [Developing Brain] - <https://europepmc.org/article/pmc/pmc4714325>
 - [Brain, bipolar] - <https://pubmed.ncbi.nlm.nih.gov/22212596/>
 - Pancan-meQTL database – [Cancer] <https://bigd.big.ac.cn/databasecommons/database/id/6207>
 - DNMIVD DNA methylation interactive visualization database [Cancer] - <http://119.3.41.228/dnmivd/>
 -
- Epigenome-wide association study databases
 - EWAS Catalog - <http://www.ewascatalog.org/>
 - EWAS Atlas - <https://bigd.big.ac.cn/ewas>
 - EWASdb: epigenome-wide association study database - <http://www.bioapp.org/ewasdb>
 - EWAS Data Hub - <https://bigd.big.ac.cn/ewa>
- Preprint services
 - arXiv (physics, mathematics, computer science, quantitative biology, statistics, engineering, and economics) <https://arxiv.org/>
 - bioRxiv (biology) <http://biorxiv.org/>
 - BITSS (research methodology) <https://osf.io/preprints/bits>
 - medRxiv (epidemiology, medicine) <https://www.medrxiv.org/>
 - NutriXiv (nutritional sciences) <https://osf.io/preprints/nutrixiv>
 - PsyArXiv (psychology) <https://psyarxiv.com/>
 - SocArXiv (social sciences) <https://osf.io/preprints/socarxiv>