

vision disorders, and dense genome-wide genotype information on more than 675,000 genetic markers generated using Affymetrix Axiom arrays. The cohort is ethnically diverse, with 7.5% Asian, 7% Latino, 3.5% African American, and 81% non-Hispanic white subjects. We identified a total of 2,147 AMD cases (46 Asian, 125 Latino, 11 African American, and 1,965 non-Hispanic whites) and 37,521 controls (2,013 Asian, 3,201 Latino, 1,168 African American, and 31,139 non-Hispanic whites) for analysis. Analyses were conducted separately for each race/ethnicity group. **Results:** In the largest group, non-Hispanic whites, we identified highly significant associations with variants in the CFH and HTRA1/ARMS2 gene regions, and genome-wide significant associations in the C2/CFB and C3 gene regions. **Conclusions:** These results confirm those of previous studies and demonstrate the power of the GERA cohort for combining information from electronic medical records with extensive genotype data. This approach can be applied to additional vision disorder phenotypes, including response to treatment and disease progression.

Keywords: Genetics; Vision Disorders; Age-Related Macular Degeneration
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B4-5:

Neighborhood Deprivation and Telomere Length: Preliminary Findings from the Kaiser Permanente Research Program on Genes, Environment, and Health (RPGEH)

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Background/Aims: Shortened telomeres have been associated with numerous adverse health outcomes. In addition, a number of environmental or external exposures, including smoking, air pollution and stress, have been reported to be associated with short telomeres. We sought to examine how neighborhood quality of participants in the RPGEH Genetic Epidemiology Research Study on Adult Health and Aging (GERA) cohort affected telomere length. **Methods:** The GERA cohort is a multi-ethnic cohort (average age = 63 years) of over 100,000 individuals with linked electronic medical records and questionnaire data. Telomere length was determined from a saliva sample in the Blackburn Laboratory using the novel Automated Telomere Length Analysis System (ATLAS) to handle the required high throughput processing of samples. Each sample was assayed six times using qPCR. Relative telomere length (T/S) was obtained from the initial concentrations of the sample telomere (T) with the corresponding sample reference gene (S). The distribution of (T/S) was found to be positively skewed and a log transformation was used to normalize the distribution. The final telomere length end point was the difference in adjusted means of telomere length per standard deviation unit by accounting for age and gender. The NDI is a standardized composite score of neighborhood quality derived from eight 2000 US Census data variables related to poverty/income, occupation, family structure, education and unemployment and normalized to a 100-point scale at the block-group level. **Results:** A higher NDI indicates greater neighborhood deprivation. The NDI was linked with residential address at time of sample collection. We observed a pattern of shorter telomere length with increasing level of neighborhood deprivation. The pattern persisted even after accounting for age, gender, race/ethnicity, smoking, BMI, and the presence of cardiovascular disease, diabetes and other comorbidity. **Conclusions:** Our results suggest that neighborhood can adversely impact telomere length. Future plans will be discussed.

Keywords: Telomere; Neighborhood Deprivation; Environment
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PS3-1:

Marshalling Site-Specific Data in Multi-Site Studies: Lessons from the eMERGE Network

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Background/Aims: The Electronic Medical Records and Genomics (eMERGE) Network is a national consortium of nine institutions supported by the National Human Genome Research Institute (NHGRI) to study genetic correlates of disease by pooling data from local biorepositories and electronic data ecosystems. Three HMORN sites participate. Twenty-one of >40 planned genome-wide association studies (GWAS) have been completed, including GWAS for chronic, cognitive, cardiovascular, gastro-intestinal, hemotologic, infectious and other phenotypes, without patient contact. Transportable algorithms rely entirely on structured data from the EMR and, optionally, clinical text using natural language processing (NLP). Salient themes include bioinformatics, genomic medicine, privacy, and community engagement. **Methods:** Algorithms defining phenotype cases and controls are developed iteratively at a primary site in conjunction with one or two secondary sites. SAS, KNIME, Python, and random manual review at multiple sites establish an algorithm's positive predictive value (PPV) and portability. Validated algorithms, published as site-agnostic pseudo code documents on a secure Web site are implemented with local tailoring at remaining sites; data are pooled for analysis. **Results:** Pseudo code documents are an efficient way for communicating the logic and content of phenotype algorithms across sites when data not available in multi-site standardized formats (such as the Virtual Data Warehouse) are required or data must be obtained using NLP. Iterative, random-sample chart validation is an important method for developing robust transportable algorithms. Business intelligence rules systems such as KNIME simplify implementation of complex algorithms and NLP. A Web site for sharing pseudo code and validation results aids communication. **Conclusions:** Experiences from the eMERGE network offer valuable lessons for conducting multi-site studies in the HMORN when non-VDW and/or NLP-derived data are required.

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PS3-2:

Informatics Challenges to Implement Pharmacogenetics to Clinical Practice

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Background/Aims: A widely-held vision arising from genomics research is to be able to use information on genomic variation to guide clinical prescriptions. This translational project is designed to test the concept that sequence information from a patient can be coupled with electronic medical records for use in personalized medicine. **Methods:** Marshfield Clinic is a health care system in Wisconsin with 2 hospitals and 52 community care centers, a member of the HMORN, and partner of the Institute for Clinical and Translational Research, University of Wisconsin – Madison. Cattails, Marshfield Clinic's internally developed outpatient EHR, has been in existence since the late 1980s. The full-feature EHR is deployed on laptop, tablet and PDA technology and supports a variety of clinical decision support (CDS) applications surrounding care management, prevention, radiology orders and medication prescribing. Much of the underlying infrastructure that supports these CDS applications will be used when integrating genomic information into clinical practice. The underlying development principles for this initiative emphasize the importance of clinical workflow, genetic result significance, and the ability to reconfigure the system as knowledge improves. **Results:** A strategy of implementing an informatics system to satisfy the needs of pharmacogenetics from both research and clinical practice was tested. One in three of the 411,851 Marshfield Clinic patients in 2011 could benefit from a pharmacogenomic test. Implementing pharmacogenetics tests of three drugs (Warfarin, Clopidogrel, and Tamoxifen) at Marshfield Clinic could prevent potential adverse events that cost \$11.3 million annually. **Conclusions:** Such an informatics system will further