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Genomic analysis of blood-mediated disorders in African Americans

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African-American populations are disproportionately affected by stroke, hypertension, skeletal diseases and renal disorders. This increased disease prevalence is thought to arise, in part, from population specific genetic variation at genes involved in disease phenotypes. We have developed a method of identifying the significant underlying genetic polymorphisms curated from National Center for Biotechnology Information (NCB) gene sets. The resulting literature curated gene sets can be used to identify modules and pathways mediating the intersection of stroke, hypertension, renal disease, and skeletal diseases.

Methods: Stroke, hypertension, renal disease, and skeletal diseases gene sets(N=2521 genes) were obtained from NCBI Gene and then projected onto the genome, annotated with their gene ontology categories and cellular pathways to draw a bioinformatics portrayal of the overlap between these diseases with disproportionate effect in African American populations. Hotspot regions were then identified in the Human genome diversity panel populations and further characterized in HapMap populations.

Results: Mapping addiction genes onto the human genome resulted in eight gene clusters, with at least 15 disease genes (Range: 15-31 genes, pV<0.005) within a 1.5Mb contiguous distance along the genome. Hotspot genes were involved in blood circulation, cell migration, and regulation of phosphorus. Analysis of Human genome diversity panel populations with Sub-Saharan African ancestry showed population differences to non-African populations.

Conclusion: We found eight regions of the genome that are strongly involved in complex disorders overrepresented in African American populations. Functional annotation of these hotspots identifies new candidate genes previously uncharacterized in the literature. Finally comparisons of polymorphism data point to 10 strong candidate variants that merit further characterization.

Biography

Fatimah Jackson received her PhD, MA, and BA (cum laude with Distinction in all Subjects) from Cornell University. She is currently Professor of Biology and Director of the W Montague Cobb Research Laboratory at Howard University and Professor Emeritus at the University of Maryland. Dr. Jackson has published extensively on population substructure in peoples of African descent, differential expressions of health disparities, and human-plant coevolution. She is the recipient of numerous national and international prizes and awards including the Norgan Award (*Annals of Human Biology*), the Ernest E. Just Prize in Medical and Public Health Research (Avery Research Institute and Medical College of South Carolina) and has been coined by Rear Admiral Dr. Helena Mishoe, National Institutes of Health, NHLBI and US Public Health Service.

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