Top Ten Things to Know
The Expressed Genome in Cardiovascular Diseases and Stroke: Refinement, Diagnosis, and Prediction

1. The human genome comprises 23 pairs of chromosomes, among which are embedded an estimated 20,000 protein-coding genes.
2. The purpose of this statement is to summarize the state-of-the-science with respect to the use of non-genetic “–omics” technologies for refinement of cardiovascular disease mechanisms.
3. Transcriptomics, epigenomics, proteomics, and metabolomics represent the analysis of macromolecule profiles that encompass different aspects of the expressed genome from DNA through cells, tissues, and organs. In addition to these “–omics” techniques, other techniques to analyze the expressed genome include peripheral cell analysis and molecular imaging.
4. Where the genome (DNA) is largely static throughout a lifestyle, “–omics” expressed genome profiles reflect the dynamic effects of the environmental influences at a given time or exposure on the genome; therefore, timing of an -omics measurement relative to disease onset is important because of the risk of reverse causation, i.e., the disease driving changes in –omics profiles rather than the –omics profiles reflecting causation of the disease.
5. There are several general considerations for assessing the expressed genome in cardiovascular disease and stroke refinement, diagnosis, and prediction including: Clinical phenotyping, refinement of mechanisms, application of new knowledge in the clinical setting, methodological readiness, and the utility of new tests in the clinical setting.
6. Transcriptomics: There are several transcriptomics techniques for measuring expression of RNA gene transcript molecules including real-time or quantitative polymerase chain reaction (PCR), microarray, and next-generation sequencing (NGS). These techniques can measure differences in gene expression to analyze the effects of an exposure, a risk factor, or a disease condition.
7. Epigenomics: Epigenomics assays such as measuring methylation at certain sites can also be done in a genome wide array to analyze thousands of potential methylation sites in genomic DNA in peripheral blood samples, for example, to identify candidate genes that may be associated with cardiovascular risk factors or outcomes.
8. Proteomics: Proteomics studies use multiple analytical techniques to assess differences in proteins expression in cells, tissues, or other biological samples. Established cohorts with clinical tissue repositories from control and affected participants offer opportunities to analyze differential protein expression in large populations which may help to identify novel biomarkers in cardiovascular diseases.
9. Metabolomics: Metabolomics, or metabolic profiling, is an analysis of low-molecular weight molecules (“metabolites”) in tissue samples used to objectively analyze biochemical changes in response to environmental or other exposures. Several analytical techniques can be used to measure changes in metabolites, including nuclear magnetic resonance (NMR) and mass-spectrometry (MS).
10. Coupled with the information that the cardiovascular community has gained from the Human Genome Project, analyzing the expressed genome using “–Omics” techniques has the potential to improve the ability to diagnose, predict, and prognosticate diseases in individual patients—a key goal of precision medicine.