Top Ten Things To Know
Genetics and Genomics for the Prevention and Treatment of Cardiovascular Disease

1. Advances in genetics technologies, including genome-wide association studies (GWASs) and whole genome sequencing, have enabled the identification of novel genomic determinants of CVDs.

2. Although increased understanding of Mendelian cardiovascular diseases (CVDs) have led to clinically useful diagnostic tests, progress in genetic testing for complex CVDs has been limited by the relatively small impact of individual variants.

3. GWASs and complementary research have revealed a number of loci associated with coronary artery disease, such as 9p21, which has also been linked to a range of related conditions and traits, including abdominal aortic and intracranial aneurysms, platelet reactivity, and stroke.

4. Recent evidence indicates that gene expression profiling may be able to distinguish ischemic stroke from other conditions, and also provide insight into the mechanism of ischemic stroke.

5. Adverse reactions to drugs such as abacavir, clopidogrel, and warfarin have been linked to specific genetic variants that may inform clinical decision making.

6. The idea that most Mendelian diseases are “rare” should be reconsidered, since the population-wide impact of these genetic variants may be greater than previously thought.

7. Translational research is most effective when performed by teams including scientists, clinician investigators, practitioners, genetic counselors, and experts in epidemiology, informatics, regulatory science, ethics, and health services research.

8. While studies have shown the ability of a multilocus genetic risk score based on validated loci to identify people at higher risk of CVD, they have yet to be integrated into clinical practice since they have only demonstrated limited clinical utility at this time.

9. Most validated GWAS variants have been identified in studies of European populations; it is critical that future large-scale genomic studies include non-European populations for both gene discovery and the optimization of risk prediction algorithms for each major ethnic group.

10. As whole genome sequencing expands into large population cohorts, major research focuses will include mining these vast data sets and increasing functional understanding as a result of progress in other “omics” fields.


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