Top Ten Things To Know
Genetics and Cardiovascular Disease

1. This paper summarizes the existing regulatory landscape with respect to the use of genetic information in clinical medicine and offers new policy recommendations designed to facilitate the safe incorporation of the latest technologies and research findings into the clinical domain.

2. The ready availability of human genetic data represents a great opportunity to improve human health by personalizing health care and has the potential to entirely transform how we think about predisposition to disease. Such advances also create new moral, ethical and legal challenges that must be addressed before we can fully realize their positive impact on human health.

3. Although recent legislation protects individuals from discrimination by employers or health insurance providers based on their genetic information, important areas of potential discrimination such as life insurance are not included. Legislation should be formulated to provide broader protection.

4. Further patenting of DNA sequences should not be approved where the ‘invention’ involves the observation of functionally unaltered human DNA because allowing these patents can lead to a monopoly on testing related to these genes, reduce access to testing, and further inhibit scientific discovery.

5. All genetic tests, including laboratory-developed genetic tests, should undergo independent review to confirm their analytic and clinical validity. An appropriate body to carry out this review would be the Food and Drug Administration. Detailed information should be made available to healthcare professionals and the public at large.

6. Sequencing based genetic testing for Mendelian cardiovascular disease provides important information for diagnosis, family screening and in some cases, therapy. Genetic testing should be carried out in a specialist center where genetic counseling is available.

7. Pharmacogenomics can be used to predict drug efficacy, adverse events, or to identify optimal doses for individual patients. Several cardiovascular pharmacogenomic tests are currently in clinical practice.

8. Genetic testing for complex cardiovascular disease such as coronary heart disease may provide additional discriminatory value in risk estimation. More data is required on the use of this information in clinical practice.

9. Genetics and genomics should be a fundamental part of the training curriculum for all health professionals.

10. It is imperative that there is significant funding for research on the genetics of cardiovascular disease, by the National Institutes of Health and other funding agencies, to promote discovery, improve assessment of variant pathogenicity, refine genotype-phenotype correlations, and gain the necessary insights into disease pathogenesis that will ultimately allow transformation of the clinical management of inherited cardiovascular disease.


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