Editor's Page

Genomics in Cardiovascular Disease: Is It Time for Population Screening?

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It is now widely known that family history of cardiovascular disease is one of the key determinants of cardiovascular risk in the general population. However, in view of the complex pathophysiology of cardiovascular diseases, searching for genetic determinants able to predict cardiovascular risk is one of the greatest challenges for researchers in the field. The ability to identify subjects in their very early life (i.e. young children) who are at high risk of developing cardiovascular disease in their far future, would be a powerful tool in the hands of clinical cardiologists, assisting them to prevent the onset or to delay the progression of cardiovascular disease.

Further to the identification of these high-risk subjects based on their demographic and family characteristics, cardiology research has been searching for specific genetic "traces" that are able to distinguish these subjects more accurately. The search for genetic determinants with significance for cardiovascular risk assessment was initially focused on identifying specific single nucleotide polymorphisms (SNPs) in key genes known to express proteins that play an important role in the development of risk factors, or which are directly involved in atherogenesis. Indeed, at least 800 SNPs in more than 90 different genes have been associated with cardiovascular risk. SNPs in a number of genes that regulate lipid and lipoprotein synthesis and metabolism have been associated with a higher incidence of dyslipidaemia, leading, at a later stage, to the development of coronary atherosclerosis.

Moreover, new genomic-based areas of therapeutics, called "pharmacogenomics" and "nutriogenomics", are being developed. Identifying patients with specific SNPs leading either to hypo-expression of beneficial anti-atherogenic, or hyper-expression of pro-atherogenic genes, may lead to genetically guided pharmacological approaches or modifications of the individuals' dietary habits. In addition to the area of pharmacogenomics, nutriogenomics is a new field of extensive research. Since SNPs are an integral component of the evolutionary process, which have resulted from the interaction between the environment and the human genome over millennia, it is likely that relatively recent changes in diet have upset this interaction with respect to the nutritional environment. It may become possible in the future to describe at least some dietary habits as "incompatible" with the individual's genetic background.

The next level in the field of cardiovascular genomics, taking into account the complexity of current genomics, is now focused on the identification of specific haplotypes, consisting of a number of individual SNPs, that are implicated in cardiovascular disease. In the hands of cardiovascular geneticists and researchers, this new, powerful tool may prove to be of much greater importance in identifying high-risk individuals and designing individual, specific, pharmacogenomic therapeutic strategies in the future than the SNP approach that is widely used today. Therefore, it is more than likely that population screening for suspect disease-related SNPs (or haplotypes) will be applied in the next few decades, aiming to prevent cardiovascular disease. Although the existing data are insufficient to support this approach in the near future, the knowledge of the structure of the human genome, along with the large amount of information generated every day and the development of new, sophisticated equipment available in many genomic centers, may bring genomics into daily clinical practice in the years ahead.