

Editorial





# Challenges in launching genomic medicine programs

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#### **Editorial**

From monogenetic diseases such as cystic fibrosis to complex ones like coronary artery disease or hypertension, to new devices and gene therapy, a look at medical innovations that are to transform the way we predict, prevent, diagnose, fight and even feel diseases. This editorial comes at a time when a dramatic increased understanding of genetic principles has led many to hopefulness that major advances in healthcare will be possible over the next years by tailoring treatment and medicines to an individual's genetic profile. Genetics was established as a major discipline in the first half of the twentieth century, though there was no or very little interest in the medical applications of genetics at the time. A crucial paradigm shift in the biomedical research field happened when in the late 1950s, genetics became an increasingly important part of medical research and practice. The completion of a high-quality, comprehensive sequence of the human genome in 2001 was a landmark event in genetic and genomic medicine. The completion of the human genome project, an international effort to map and sequence all the human genes and marked the beginning of the "Genomic Era". Some reflected that sequencing the human genome was only "the end of the beginning," as attention turned to the monumental task of discovering how the genes interact with each other and with environmental factors. The project's ground breaking research strategies and experimental technologies have generated an even larger and more complex amount of genomic data sets that have poured into public databases, transforming the study of virtually all life processes.

Our growing understanding of the workings of the human genome is posing a new challenge: How to use that data to change the course of diseases. Soon, it was clear that post-genomic technology could make it possible to obtain detailed profiles of specific genes and their interactions, involved in complex diseases and their link to drug action. This would ultimately lead to an understanding of individual variation in response to a wide range of therapeutic agents. The next challenge is to go from genomics to proteomics and from proteome to pathology and thus the "Post Genomic and Personalized Medicine Era" began. Today, one of the major challenges is interpreting the polygenic and multifactorial etiology of common diseases, such as cancer, cardiovascular, allergic, auto-immune and degenerative disorders. Although genomic medicine is early in its development, some areas where it has shown promise include: Risk assessment; early detection; diagnosis; prognosis; tailored treatment and Pharmacogenomics. The integration of new genetic technologies into clinical practice has found multiple challenges and barriers, but still holds great promise for the personalization of medical care and genomic medicine, particularly the use of large-scale DNA sequencing for genome-wide genetic testing. One of these challenges may be modest to low appreciation by clinicians, health-care institutions and payers of the potential for genomics to improve patient care, as the evidence of clinical utility is limited at present time. However, several academic medical centers and integrated health systems are actively implementing programs for applying genomic medicine (the use an individual's genotypic

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information in the clinical care). The National Human Genome Research Institute, in collaboration with several other National Institutes of Health is currently exploring approaches for facilitating access to the research needed to implement genomic medicine on an ever-widening scale. The efforts invested in genomic research by both public and private sector stake holders have produced crucial information about the role of genetic variants in health and disease.

The progress derived from genomics to increase patients' health care is enhanced by different areas of research including: bio-informatics, proteomics and important social and public diffusion and education to help the public understand the state of the genomic art. For many genomic applications that are proven to bevalid and utile, there may be limited adoption and implementation, suggesting there are other barriers, some of them being cost concerns and scarce data availability, which must be overcomes clinical practice is changed and genomic medicine is adopted into clinical care.

#### **Conclusion**

Genomic technologies are changing fast and now we all understand that for new genomic technologies to be utilized successfully in medicine, programs must quantitatively and qualitatively address concerns in health care Institutions. The path from a promising discovery to an effective treatment often takes a decade or more, however, it is difficult to predict how quickly new technologies will become adopted in routine clinical practice. It seems likely that it will occur more rapidly than typically observed. It is probable that, in less than 10 years, we will reach a point where the analytic validity of sequencing technologies are high, making it easy to access and clinically interpret genomic information. It is now time to advance and ensure that basic and clinical research genetic results are applied for the benefit of patients by structuring genomic medicine implementation programs.

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### **Conflict of interest**

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