

# Mayo Clinic Proceedings

## Editorial

### The Genomic Revolution and Medicine

Completion of the Human Genome Project represents the first step in a process that will revolutionize medicine in the 21st century. The “race to the finish line” between the “public” and “private” Human Genome Project depicted in the popular press was really a race to the starting line with regard to the application of genomics to medicine.

Research activities downstream from the early, descriptive portions of the Human Genome Project promise to fundamentally alter the way in which future generations of health care professionals will diagnose, treat, and—ultimately—prevent disease. Unlike the early experiences with genetic medicine, in which gross aberrations of genetic material and their influence on relatively small populations of patients were typically studied, the new genomic revolution has the promise to better explain the pathomechanisms underlying highly prevalent illnesses such as coronary artery disease, cancer, and neurodegenerative disorders. Improved understanding of the multiple, interacting genetic components of disease pathophysiology should make it possible to design specific, “targeted” therapies. Furthermore, it should be possible to “individualize” drug and other treatments to accommodate both inherited differences in the pathophysiology of a single patient’s disease process and genetic variation in that patient’s ability to metabolize drugs. These issues are addressed by the field of “pharmacogenomics.” Achieving the promise of the genomic revolution for medicine will also require the development of new clinical laboratory tests, including those that use microarrays (chips), that in turn can be used for DNA sequence analysis and for evaluation

of gene expression in normal and diseased tissue. Because genes encode proteins, the genomic revolution has also led to the development of “proteomics.” Proteomics involves measurement not of single proteins, but rather of all the proteins expressed in a cell or tissue.

The bounty of information resulting from the development of genomic and proteomic science has required the parallel development of computer-based techniques for managing vast collections of data and, even more important, providing ready access to those data by medical scientists and practitioners. In response, the discipline of bioinformatics has developed new techniques for organizing, storing, and analyzing the acquired information and, in many instances, disseminating the information via the World Wide Web. Although the promise of the genomic revolution for medicine is truly unprecedented, it is important that enthusiasm for the science be tempered by sensitivity to public concerns regarding the potential misuse of genomic information. The possibility of loss of confidentiality, issues of group stigmatization, and the difficult issue of genomic predestination must all be addressed if humankind is to reap the full benefits of the potential of this exciting new science.

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**See also pages 773 and 785.**

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As is readily apparent, evolving genomic science has associated concepts and a vocabulary that may challenge the student of medicine and excite or frustrate the mid-career practitioner. The scope of genomic knowledge and the applications are so broad that it is difficult to envision a disease process, patient, or clinician who will not be affected in the future. It is important that graduate, postgraduate, and continuing medical education keep abreast of the new information so that new discoveries can be translated into clinical practice rapidly and appropriately.

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Individual reprints of this editorial are not available. The entire Primer on Medical Genomics will be available for purchase from the *Proceedings* Editorial Office at a later date.

In recognition of this challenge, the Mayo Clinic Genomics Education Steering Committee, composed of physicians, scientists, and other professionals, working in concert with the editorial board of *Mayo Clinic Proceedings*, has prepared a series of articles to help *Proceedings* readers understand and appreciate the genomics-related changes in medicine that are under way. Published as a new section of the *Proceedings* entitled "Medical Genomics," care is taken in each article to represent the underlying science fairly while effectively communicating the material to the medical practitioner, regardless of the stage of a career. Through these articles, the clinician will become familiar with the vocabulary of genomics, proteomics, and bioinformatics, the current status of the integration of genomics and proteomics into medicine, and the "tools" needed to make that integration possible. In addition, the social and ethical issues of genetic testing, known as *genoethics*, will be explored.

Only once in the history of our species will humankind experience the process of seminal discovery, and participate in the initial clinical applications, resulting from the genomic revolution. All of us involved in medicine can only be grateful that we have the opportunity to be present

at the creation of this science and can participate in the clinical and educational experiences it affords.

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