

# Chipping away at genomic medicine

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The National Human Genome Research Institute of the National Institutes of Health is pleased to sponsor this special supplement of *Nature Genetics*. Every area of biology has been revolutionized by the vast increase in the availability of the DNA sequence of several genomes. With this explosion in sequence-based information has come an insatiable need for computational methods to view and to use this information so that the genomic issues related to medicine can begin to be addressed.

The overwhelming focus of scientists for years has been on collecting and disseminating DNA sequence information through efforts such as the Human Genome Project. Parallel to this, remarkable progress has been made in the development of numerous technologies and tools for assessing global alterations in gene expression, in particular the differences between normal and disease samples. Most of the *Chipping Forecast II* is devoted to articles describing how such technical developments and 'global gene profiling' can provide a powerful research tool and can also have concrete and significant implications for introducing genomic techniques into medical practice.

An equally outstanding component of this supplement discusses the many different goals addressed by these high-throughput genomic technologies. Such goals range from hypothesis-generating attempts to assign probable functions to newly identified genes (for example, by comparison with the expression patterns of known genes) to the identification of therapeutic targets and the delineation of complex patterns of gene expression that provide a potentially 'pathogenomic' molecular phenotype. But the use of these technologies is not restricted to basic biology. They are finding applications in areas

as diverse as, for example, the identification of genetic signatures of pathogen exposure and toxicogenomics (the study of the genetic basis of an individual's response to environmental factors such as drugs and pollutants).

In addition, an important element of this supplement addresses commonly used approaches for data collection and analysis, and discusses the introduction of universal standards for contextual validation. The application of these very powerful methods is often limited owing to a lack of rigorous standards for data collection, analysis and validation. One of the key factors that will ultimately decide the future success of applying microarrays to biological and clinical issues is standardization, not only in an experimental sense but also in a computational sense. Significant advances have been made toward introducing criteria for describing how a particular microarray experiment has been carried out and for representing the data derived from that experiment. The efforts of many in the microarray community are starting to push the field toward a point where scientists will be able to accurately compare results from different laboratories—a comparison that so far has been problematic.

Genomics is being increasingly defined as the comprehensive study of whole sets of genes, gene products and their interactions. As such, the *Chipping Forecast II* highlights several tools that will facilitate the global and high-throughput analysis of genes and gene products. We trust that it will serve as a timely introduction to the subject for basic investigators, physicians and medical scientists interested in functional genomics, mRNA profiling and proteomics, and in the applications of high-throughput technologies.

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