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Australia: Melbourne, Australia Tel: (03) 9212-8585 Fax: (03) 9212-8502

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GENOME RESEARCH

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Cold Spring Harbor, New York 11724
Phone (516) 367-8492
Fax (516) 367-8334
<http://www.cshl.org>

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COVER Sequence and schematic representation of 130-kb sequence contig from the IDS region. (For details, see Timms et al., p. 71.)

Genome Research and



Despite its youth, the Genome Project is now a critical presence in biomedical research. The goals of mapping and sequencing the genomes of the human and several model organisms were initially endorsed mainly by geneticists, who foresaw in the maps and sequence an opportunity to address problems that were largely the "stuff" of conjecture. When successful, geneticists argued, genes would no longer remain the intangible factors inferred by rules of inheritance in crosses or human pedigrees but would be the currency of the new genetics. Human genetics, in particular, would be revolutionized, since for the first time relevant questions about human biology could be answered directly in the human rather than by extrapolation from other organisms.

The Genome Project has been wildly successful. It has produced unprecedented amounts of genetic information about organisms at all taxonomic levels and has catalyzed the development of technology for the benefit of research in diverse scientific disciplines. Several areas are particularly notable. Comprehensive genetic maps with highly polymorphic DNA markers have been completed for the human and mouse genomes. These maps and markers have revolutionized the way that phenotypes are now connected to their underlying genes. Indeed, the identification of genetic linkage in these organisms is now routine for single gene traits and increasingly being extended to the study of quantitative trait loci and complex diseases. To complement these efforts, first-generation physical maps of human chromosomes are approaching completion. These maps, along with cloned DNA reagents, have improved our ability to identify genes associated with human disease on the basis of their chromosomal location (i.e., positional and positional candidate cloning), and indeed new disease genes are being identified at an impressive rate. These successes in turn have led to the discovery of novel molecular mechanisms that

cause genetic disease, such as dynamic mutation and imprinting, and critical features of genome structure that explain its underlying function.

Equally exciting is the progress in the maps and sequence from many key organisms in experimental biology. The entire genomic sequences of several important bacterial species have been finished, and completion of the *Saccharomyces cerevisiae* genome sequence is imminent. Many megabases of genomic sequence are already completed for the nematode *Caenorhabditis elegans*, and an analogous effort for the *Drosophila* genome is well underway. In addition, enormous amounts of data on human expressed sequences have been generated. While these past accomplishments are impressive, it is perhaps more exciting that the Genome Project will continue to make profound advances in data acquisition and technology development, and indeed, is poised to begin an almost explosive phase of DNA sequencing. For the first time, the complete human genome sequence appears within our reach. The databases that maintain these maps and sequences are constantly evolving to keep pace with the flood of new data, thereby facilitating access to the information by the scientific community.

A founding principle of the Genome Project is that the generation of maps, sequences, and associated reagents for several organisms would allow more efficient progress from the search for a gene to the study of its function. Indeed, there is mounting evidence that such a paradigm shift is occurring. This is because researchers in all areas of biology are learning how to find and use genomic data to enhance their studies. Just as the double-helical structure of DNA made obvious some of its properties, it is anticipated that knowledge of the genome structure and sequence will begin to reveal its function. While the sequence will itself be new, our challenge will be to formulate and answer previously intractable biological questions.

The success of the Genome Project, and the genetic discoveries it has made possible, has spawned a discipline that can be dubbed Genome Research. To capture the exciting advances in this rapidly growing field and assist in their rapid transmission to the scientific community, we are launching this journal, *Genome Research*. In it we wish to publish the best and most creative research being performed on physical and genetic mapping, DNA sequencing, gene discovery, informatics, statistical and mathematical methods, DNA-based technology development, gene function, genome structure and function, and human disease. Because the field demands ever-increasing sophistication in information handling and analysis, the journal will have a strong electronic presence, which will include an associated World Wide Web site for the distribution of data and other information that will supplement the material published in print. Since in the future genetic information will influence the lives of citizens, *Genome Research* will also publish discussions about the new ethical and legal issues that inevitably will arise and require careful thought for their solution.

It is a quirk of history, but perhaps fitting, that the official end of the Genome Project will come soon after we enter the next century. The fruits of this labor will be our legacy and, in the next century, the genomic tools and reagents we have developed will allow us to probe biology in an unprecedented manner. It will take all of the scientific curiosity and creativity that we can muster. This journal—*Genome Research*—will be about that future.

Aravinda Chakravarti

Richard Gibbs

Eric Green

Richard Myers

Editors, *Genome Research*