A scientific milestone

Scientists unravel genetic code for human chromosome 22

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The publication of the complete genetic code of the human chromosome 22 in the December 2 issue of the scientific journal *Nature* is an important scientific achievement, which has enormous potential for medical science and the study of human developmental biology and evolution. It is the first human chromosome to be mapped by the Human Genome Project and involved the collaborative efforts of over 200 scientists from the United Kingdom, Japan, the United States, Canada and Sweden.

The Human Genome Project has undertaken the sequencing of the entire human genome—that is, the complete set of human chromosomes—and estimates that the task will be completed by 2003.

Dr. Ian Dunham at the Sanger Center at Cambridge in England, who headed the research team said that "this is the first time we've really been able to see a whole human chromosome in all its detail—the organisation of all of the genes and how they relate to each other." Dr. Dunham also commented on the significance of the international collaboration: "It is that strength which has let us to be able to reach this milestone."

Professor of Molecular Medicine at Hammersmith Hospital in London, James Scott described the publication as "a fantastic resource for looking at human population genetics and disease gene susceptibility in a way that has just not been really possible until now."

The genetic map is being published on the Internet and updated daily, as new sections of the genome are sequenced for the use of the international scientific community. In fact, a paper copy of the entire genome will probably never be published, as it would take at least half a million pages to print.

The human genetic code is found in structures contained within the nucleus of each cell known as chromosomes. Humans have 23 pairs of chromosomes, which contain the genetic instructions for the human body. Chromosomes consist of tightly coiled strands of the complex chemical deoxyribonucleic acid (DNA). DNA is made up of two inter linked double helix chains of four chemical bases—adenine, thymine, guanine, and cytosine. The sequence of base pairs on the two chains makes up the genetic code. The scientists have unscrambled this code for chromosome 22.

The genetic sequence of the chromosome was determined by cutting it up into sections, each of about 500 bases, which were then inserted into the chromosome of either a yeast or bacteria cell. The microorganism replicated the piece of DNA until the scientists had sufficient quantities for sequencing. A sophisticated computer model was then used to determine the order of the pieces on the chromosome and to identify any genes.

Even though chromosome 22 is the second smallest human chromosome, scientists had to sequence 33 million DNA bases. The chromosome is known to contain at least 545 genes and possibly as many as 1,000. The precise function of most of the genes is still unknown.

The publication represents an important step forward for medical science. The chromosome has been linked to 27 genetic diseases, which are caused by defective genes on the chromosome. These include schizophrenia, chronic myeloid leukaemia and trisomy 22, the second most common cause of miscarriages. The chromosome has been implicated in a number of

cancers as well as disorders of the nervous system and foetal development.

Scientists will be able to examine the evolution of the human species in greater detail by comparing the structure of genes and the chromosome itself in various organisms and examining how they have changed over evolutionary history. Already one gene on chromosome 22 has been found virtually unchanged in primitive yeast cells. Why the structure of the gene should be preserved over millions of years may provide important clues for evolutionary scientists.

The new genetic map also provides developmental biologists with a valuable tool. For the first time, they will be able to follow human development from the level of the gene to the formation of proteins, how these proteins interact to form cells and ultimately a complete human being. Researchers will be able to study how medical disorders develop at a molecular level and to devise techniques to prevent the diseases developing in the first place.

The sequencing of chromosome 22 provides an indication of the enormous scientific potential of this genetic research. The completion of the Human Genome Project, providing a comprehensive map of human genes, will certainly mark a key scientific turning point and provide fresh impetus to what is already a profound revolution in human biology and medical science.