

Working draft of the human genome completed

Both the public Human Genome Project and biotechnology company Celera Genomics have put together their first compilations of the human genome DNA nucleotide sequences. After two years of often heated competition, the leaders of the public and private sector efforts met in June in Washington to praise each other's achievements and announce the historic milestone. James Watson, Director of the Cold Spring Harbor Laboratory, commented wryly: "From the public viewpoint, everyone has gained".

The Human Genome Project working draft covers 97% of the human genome, and a final, completed version is now expected within the next two years, with work centering on filling the gaps in the draft sequence and raising overall accuracy to 99.99%. Scientists at 16 institutions in France, Germany, Japan, the People's Republic of China, the United Kingdom and the United States generated around 82% of the sequencing data in the public project which made its results available without delay via the Internet.

The coordinated work of unlocking the secrets of DNA, the double-stranded molecule packaged into 23 chromosomes which may code for up to 150 000 genes, began in 1990. The goal of the human genome research has been to obtain a single reference sequence of the three billion chemical bases that make up human DNA. But although the working draft is a major milestone, therapeutic applications of the technology are only in their infancy.

The aim now is to use the genetic blueprint to match genes with their functions and to detect genetic variations. Already, dozens of disease genes have been pinpointed by researchers accessing the public working draft. More than 300 000 individual genetic variations — single nucleotide polymorphisms or SNPs — have been found which are crucial to the study and understanding of human diseases and disorders.

Identifying genes and mutated genes important in the development of disease will also lead to better-targeted medicines. Latest predictions are that new medicines generated from the project will reach the market in large numbers within the next 10 to 15 years.

Research is on-going to identify the genes and genetic variations that alter the risk of developing a range of common diseases, including Alzheimer disease, asthma, different forms of cancer, and Parkinson disease. Individual genetic data, which may one day be carried embedded in personal smart cards, will mean that drugs can be tailor-made for each patient, dramatically reducing the risk of side effects.

The technology will also lead to better screening for diseases, but that will raise ethical issues affecting insurance, employment and even marriage. Controversies over patenting, as already seen with the hereditary breast cancer genes, are also likely to become a growing problem.

The total costs of producing the public project working draft, which involved input from 1000 scientists around the world, is estimated at US\$ 300 million. Work on sequencing began in 1990 and the total amount spent has been estimated at US\$ 3 billion. However, that includes funding for projects as diverse as developing computer methods to analyse genomes to studies looking at the ethical, legal and social issues surrounding genetics. ■

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