

Human Genome Project



Yesterday

- Just a half-century ago, very little was known about the genetic factors that contribute to human disease.
- In 1953, James Watson and Francis Crick described the double helix structure of deoxyribonucleic acid (DNA), the chemical compound that contains the genetic instructions for building, running and maintaining living organisms.
- Methods to determine the order, or sequence, of the chemical letters in DNA were developed in the mid-1970s.
- In 1990, the National Institutes of Health (NIH) and the Department of Energy joined with international partners in a quest to sequence all 3 billion letters, or base pairs, in the human genome, which is the complete set of DNA in the human body. This concerted, public effort was the Human Genome Project.
- The Human Genome Project's goal was to provide researchers with powerful tools to understand the genetic factors in human disease, paving the way for new strategies for their diagnosis, treatment and prevention.
- From the start, the Human Genome Project supported an Ethical, Legal and Social Implications research program to address the many complex issues that might arise from this science.
- All data generated by the Human Genome Project were made freely and rapidly available on the Internet, serving to accelerate the pace of medical discovery around the globe.
- The Human Genome project spurred a revolution in biotechnology innovation around the world and played a key role in making the U.S. the global leader in the new biotechnology sector.
- In April 2003, researchers successfully completed the Human Genome Project, under budget and more than two years ahead of schedule.

Today

- The Human Genome Project has already fueled the discovery of more than 1,800 disease genes.
- As a result of the Human Genome Project, today's researchers can find a gene suspected of causing an inherited disease in a matter of days, rather than the years it took before the genome sequence was in hand.
- There are now more than 2,000 genetic tests for human conditions. These tests enable patients to learn their genetic risks for disease and also help healthcare professionals to diagnose disease.
- At least 350 biotechnology-based products resulting from the Human Genome Project are currently in clinical trials.
- Having the complete sequence of the human genome is similar to having all the pages of a manual needed to make the human body. The challenge now is to determine how to read the contents of these pages and understand how all of these many, complex parts work together in human health and disease.
- One major step toward such comprehensive understanding was the development in 2005 of the HapMap (<http://hapmap.ncbi.nlm.nih.gov/>), which is a catalog of common genetic variation, or haplotypes, in the human genome. In 2010, the third phase of the HapMap project was published, with data from 11 global populations, the largest survey of human genetic variation performed to date. HapMap data have accelerated the search for genes involved in common human diseases, and have already yielded impressive results in finding genetic factors involved in conditions ranging from age-related blindness to obesity.
- The tools created through the Human Genome Project continue to underlie efforts to characterize the genomes of important organisms used extensively in biomedical research, including fruit flies, roundworms, and mice.

- NIH's Ethical, Legal and Social Implications program has become a model for other research efforts seeking to address ethical issues in a proactive manner (<http://www.genome.gov/10001618>).
- With the drastic decline in the cost of sequencing whole exomes or genomes, groundbreaking comparative genomic studies are now identifying the causes of rare diseases such as Kabuki and Miller syndromes.
- Much work still remains to be done. Despite many important genetic discoveries, the genetics of complex diseases such as heart disease are still far from clear.
- Pharmacogenomics is a field that looks at how genetic variation affects an individual's response to a drug. Pharmacogenomic tests can already identify whether or not a breast cancer patient will respond to the drug Herceptin, whether an AIDS patient should take the drug Abacavir, or what the correct dose of the blood-thinner Warfarin should be.

Tomorrow

- An ambitious new initiative, The Cancer Genome Atlas (<http://cancergenome.nih.gov/>), aims to identify all the genetic abnormalities seen in 50 major types of cancer.
- Based on a deeper understanding of disease at the genomic level, we will see a whole new generation of targeted interventions, many of which will be drugs that are much more effective and cause fewer side effects than those available today.
- NIH-supported access to high-throughput screening of small molecule libraries will provide academic researchers with powerful new research probes to explore the hundreds of thousands of proteins believed to be encoded by the approximately 25,000 genes in the human genome, and will provide innovative techniques to spur development of new, more effective, types of drugs.
- NIH is striving to cut the cost of sequencing an individual's genome to \$1,000 or less. Having one's complete genome sequence will make it easier to diagnose, manage and treat many diseases.
- Individualized analysis based on each person's genome will lead to a powerful form of preventive, personalized and preemptive medicine. By tailoring recommendations to each person's DNA, health care professionals will be able to work with individuals to focus efforts on the specific strategies — from diet to high-tech medical surveillance — that are most likely to maintain health for that particular individual.
- The increasing ability to connect DNA variation with non-medical conditions, such as intelligence and personality traits, will challenge society, making the role of ethical, legal and social implications research more important than ever.

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