Genetics Home Reference
Your Guide to Understanding Genetic Conditions

Handbook

Help Me Understand Genetics

The Human Genome Project and Genomic Research

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Chapter 7

The Human Genome Project and Genomic Research

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What is a genome?

A genome is an organism's complete set of DNA, including all of its genes. Each genome contains all of the information needed to build and maintain that organism. In humans, a copy of the entire genome—more than 3 billion DNA base pairs—is contained in all cells that have a nucleus.

For more information about genomes:

The U.S. Department of Energy Office of Science provides background information about the human genome in its fact sheet The Science Behind the Human Genome Project (http://www.ornl.gov/TechResources/Human_Genome/project/info.html).

The NCBI Science Primer (http://www.ncbi.nlm.nih.gov/About/primer/genetics_genome.html) offers more detailed information about the structure and function of the human genome under the heading "What Is a Genome?"

What was the Human Genome Project and why has it been important?

The Human Genome Project was an international research effort to determine the sequence of the human genome and identify the genes that it contains. The Project was coordinated by the National Institutes of Health and the U.S. Department of Energy. Additional contributors included universities across the United States and international partners in the United Kingdom, France, Germany, Japan, and China. The Human Genome Project formally began in 1990 and was completed in 2003, 2 years ahead of its original schedule.

The work of the Human Genome Project has allowed researchers to begin to understand the blueprint for building a person. As researchers learn more about the functions of genes and proteins, this knowledge will have a major impact in the fields of medicine, biotechnology, and the life sciences.

For more information about the Human Genome Project:

The National Human Genome Research Institute offers a fact sheet about the Human Genome Project (http://www.genome.gov/10001772) and a list of frequently asked questions (http://www.genome.gov/11006943).

A brief description of the Project and links to many additional resources are available from the Human Genome Project Information web site (http://www.ornl.gov/TechResources/Human_Genome/home.html), a service of the U.S. Department of Energy Office of Science.

The U.S. Department of Energy Office of Science also provides a fact sheet called Potential Benefits of Human Genome Project Research (http://www.ornl.gov/sci/techresources/Human_Genome/project/benefits.shtml).

What were the goals of the Human Genome Project?

The main goals of the Human Genome Project were to provide a complete and accurate sequence of the 3 billion DNA base pairs that make up the human genome and to find all of the estimated 30,000 to 40,000 human genes. The Project also aimed to sequence the genomes of several other organisms that are important to medical research, such as the mouse and the fruit fly.

In addition to sequencing DNA, the Human Genome Project sought to develop new tools to obtain and analyze the data and to make this information widely available. Also, because advances in genetics have consequences for individuals and society, the Human Genome Project committed to exploring the consequences of genomic research through its Ethical, Legal, and Social Implications (ELSI) program.

For more information about the Human Genome Project's goals:

The U.S. Department of Energy Office of Science offers an overview of the Human Genome Project's 5-year goals (http://www.ornl.gov/TechResources/Human_Genome/hg5yp/), including a table outlining the goals and when they were achieved.

The National Human Genome Research Institute provides a fact sheet about DNA sequencing (http://www.genome.gov/10001177).

What did the Human Genome Project accomplish?

In April 2003, researchers announced that the Human Genome Project had completed a high-quality sequence of essentially the entire human genome. This sequence closed the gaps from a working draft of the genome, which was published in 2001. It also identified the locations of many human genes and provided information about their structure and organization. The Project made the sequence of the human genome and tools to analyze the data freely available via the Internet.

In addition to the human genome, the Human Genome Project sequenced the genomes of several other organisms, including brewers' yeast, the roundworm, and the fruit fly. In 2002, researchers announced that they had also completed a working draft of the mouse genome. By studying the similarities and differences between human genes and those of other organisms, researchers can discover the functions of particular genes and identify which genes are critical for life.

The Project's Ethical, Legal, and Social Implications (ELSI) program became the world's largest bioethics program and a model for other ELSI programs worldwide. For additional information about ELSI and the program's accomplishments, please refer to "What were some of the ethical, legal, and social implications addressed by the Human Genome Project?" on page 7

For more information about the accomplishments of the Human Genome Project:

An overview of the Project's accomplishments is available in the National Human Genome Research Institute press release International Consortium Completes Human Genome Project (http://www.genome.gov/11006929).

The U.S. Department of Energy Office of Science provides links to information about the Project's activities as part of its fact sheet Human Genome Project Completion: 1990-2003 (http://www.ornl.gov/TechResources/Human_Genome/project/50yr.html).

The complete sequence of the human genome and articles analyzing the sequence were published in early 2003. The Human Genome Project Information web site provides an index of these landmark scientific papers (http://www.ornl.gov/TechResources/Human_Genome/project/journals/journals.html).

What were some of the ethical, legal, and social implications addressed by the Human Genome Project?

The Ethical, Legal, and Social Implications (ELSI) program was founded in 1990 as an integral part of the Human Genome Project. The mission of the ELSI program was to identify and address issues raised by genomic research that would affect individuals, families, and society. A percentage of the Human Genome Project budget at the National Institutes of Health and the U.S. Department of Energy was devoted to ELSI research.

The ELSI program focused on the possible consequences of genomic research in four main areas:

- Privacy and fairness in the use of genetic information, including the potential for genetic discrimination in employment and insurance.
- The integration of new genetic technologies, such as genetic testing, into the practice of clinical medicine.
- Ethical issues surrounding the design and conduct of genetic research with people, including the process of informed consent.
- The education of healthcare professionals, policy makers, students, and the public about genetics and the complex issues that result from genomic research.

For more information about the ELSI program:

Information about the ELSI program at the National Institutes of Health, including program goals and activities, is available in the fact sheet Ethical, Legal and Social Implications (ELSI) Research Program (http://www.genome.gov/10001618) from the National Human Genome Research Institute. The About ELSI web page (http://www.genome.gov/10001754) provides a more detailed discussion of the program.

The U.S. Department of Energy Office of Science offers two fact sheets on the ELSI program, each of which includes links to many additional resources:

Ethical, Legal, and Social Issues (http://www.ornl.gov/TechResources/Human_Genome/elsi/elsi.html)

Ethical, Legal, and Social Issues Research (http://www.ornl.gov/TechResources/Human_Genome/research/elsi.html)

What are the next steps in genomic research?

Discovering the sequence of the human genome was only the first step in understanding how the instructions coded in DNA lead to a functioning human being. The next stage of genomic research will begin to derive meaningful knowledge from the DNA sequence. Research studies that build on the work of the Human Genome Project are under way worldwide.

The objectives of continued genomic research include the following:

- Determine the function of genes and the elements that regulate genes throughout the genome.
- Find variations in the DNA sequence among people and determine their significance. These variations may one day provide information about a person's disease risk and response to certain medications.
- Discover the 3-dimensional structures of proteins and identify their functions.
- Explore how DNA and proteins interact with one another and with the environment to create complex living systems.
- Develop and apply genome-based strategies for the early detection, diagnosis, and treatment of disease.
- Sequence the genomes of other organisms, such as the rat, cow, and chimpanzee, in order to compare similar genes between species.
- Develop new technologies to study genes and DNA on a large scale and store genomic data efficiently.
- Continue to explore the ethical, legal, and social issues raised by genomic research.

For more information about the genomic research following the Human Genome Project:

The National Human Genome Research Institute supports research in many of the areas described above. The Institute provides detailed information about its research initiatives at NIH and nationwide (http://www.genome.gov/Research/). In addition, the NIH Roadmap (http://nihroadmap.nih.gov/) outlines major initiatives in biomedical research.

The U.S. Department of Energy's Genomes to Life program will use genomic data and new technologies to obtain a fundamental understanding of living systems. The

Genomes to Life web site (http://doegenomestolife.org/) offers information about the program.

The U.S. Department of Energy Office of Science provides a look at the possible benefits and applications of future research in the article Fast Forward to 2020: What to Expect in Molecular Medicine (http://www.ornl.gov/hgmis/medicine/tnty.html).

What is pharmacogenomics?

Pharmacogenomics is the study of how genes affect a person's response to drugs. This relatively new field combines pharmacology (the science of drugs) and genomics (the study of genes and their functions) to develop effective, safe medications and doses that will be tailored to a person's genetic makeup.

Many drugs that are currently available are "one size fits all," but they don't work the same way for everyone. It can be difficult to predict who will benefit from a medication, who will not respond at all, and who will experience negative side effects (called adverse drug reactions). Adverse drug reactions are a significant cause of hospitalizations and deaths in the United States. With the knowledge gained from the Human Genome Project, researchers are learning how inherited differences in genes affect the body's response to medications. These genetic differences will be used to predict whether a medication will be effective for a particular person and to help prevent adverse drug reactions.

The field of pharmacogenomics is still in its infancy. Its use is currently quite limited, but new approaches are under study in clinical trials. In the future, pharmacogenomics will allow the development of tailored drugs to treat a wide range of health problems, including cardiovascular disease, Alzheimer disease, cancer, HIV/AIDS, and asthma.

For more information about pharmacogenomics:

The U.S Department of Energy Office of Science offers a fact sheet on pharmacogenomics (http://www.ornl.gov/TechResources/Human_Genome/medicine/pharma.html). This resource outlines the anticipated benefits of this approach and lists barriers to progress.

The National Center for Biotechnology Information provides a discussion of this topic as part of its Science Primer: One Size Does Not Fit All: The Promise of Pharmacogenomics (http://www.ncbi.nlm.nih.gov/About/primer/pharm.html).

A list of clinical trials involving pharmacogenomics (http://clinicaltrials.gov/search/term=pharmacogenomics+OR+pharmacogenetics) is available from ClinicalTrials.gov, a service of the National Institutes of Health.

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