The Human Genome Project: Frequently Asked Questions

On April 14, 2003, the National Human Genome Research Institute (NHGRI), the Department of Energy (DOE) and their partners in the International Human Genome Sequencing Consortium announced the successful completion of the Human Genome Project.

What is a genome?

A genome is an organism's complete set of deoxyribonucleic acid (DNA), a chemical compound that contains the genetic instructions needed to develop and direct the biological activities of every organism. DNA molecules are made of two twisting, paired strands. Each strand is made of four chemical units, called nucleotide bases. The bases are adenine (A), thymine (T), guanine (G) and cytosine (C). Bases on opposite strands pair specifically: an A always pairs with a T, and a C always with a G. The human genome contains approximately 3 billion of these base pairs, which reside in the 23 pairs of chromosomes within the nucleus of all our cells. The Human Genome's Project's principal task was determining the order or sequence of those As, Ts, Cs, and Gs.

What is sequencing and how do you sequence a genome?

Sequencing means determining the exact order of the base pairs in a segment of DNA. Human chromosomes range in size from about 50 million to 300 million base pairs. One complete set of genomic instructions is about 3 billion base pairs. Specialized machines called DNA sequencers – the technologies have rapidly evolved over the last decade – use techniques of biochemistry to automatically determine the base pair order and deposit the read out in a computer.

Whose DNA was sequenced for the Human Genome Project?

This is intentionally not known to protect the privacy of the volunteers who provided DNA samples for sequencing. The sequence was derived from the DNA of several volunteers who came from Upstate New York. To ensure that the identities of the volunteers cannot be revealed, a careful process was developed to recruit the volunteers and to collect and maintain the blood samples that were the source of the DNA.

What does it mean when you say you've completed the Human Genome Project?

The main goals of the Human Genome Project included the essential completion of a high-quality version of the human sequence, the creation of physical and genetic maps of the human genome and the mapping and sequencing of a set of five model organisms, including the mouse. All of these goals were achieved ahead of schedule and under budget.

Is the human genome completely sequenced?

Yes, within the limits of today's technology, the reference human genome sequence used in genome research is as complete as it can be. Only small gaps of DNA that could not be recovered using current sequencing methods remain.

Who owns the human genome?

Every part of the genome sequenced by the Human Genome Project was made public immediately. Private companies have filed thousands of patents on human genes, including Myriad Genetics, Inc., which patented the *BRCA1* and *BRCA2* gene mutations that cause inherited forms of breast cancer. In 2013, the lawsuit over Myriad's patents headed towards a final resolution by the Supreme Court. The court's ruling in this case will set a binding precedent for the lower courts.

Who participated in the international Human Genome Project consortium?

The Human Genome Project could not have been completed as quickly or as effectively without the strong participation of international institutions. In the United States, contributors to the effort include the National Institutes of Health (NIH) and the U.S. Department of Energy (DOE). A majority of the actual genome sequencing was conducted at universities and research centers throughout the United States, the United Kingdom, France, Germany, Japan and China. For a complete list, go to http://www.genome.gov/11006939.

How much did the Human Genome Project cost U.S. taxpayers?

In 1990, Congress established funding for the Human Genome Project and set a target completion date of 2005. Although estimates suggested that the project would cost a total of $3 billion over this period, the project ended up costing NIH about $2.7 billion in FY 1991 dollars and was completed more than two years ahead of schedule. A 2011 report, updated in 2013, from the research firm Battelle Technology Partnership Practice, estimated that between 1988 and 2012, federal investment in genomic research generated nearly $1 trillion of economic output from an investment of $5.4 billion in the HGP (in 2012 dollars). This figure equates to a return on investment of 178:1, and total tax revenues from the genomics sector and suppliers of $54.8 billion by 2012.

Why was a portion of the NHGRI budget set aside for ethical considerations?

Five percent of the annual budget of the NHGRI is dedicated to examining ethical, legal and social implications (ELSI) related to human genome research, incorporating specific recommendations into the activities of NHGRI and providing guidance to policymakers and the public. NHGRI's ELSI program, within the Division of Genomics and Society, is considered unprecedented in biomedical science in terms of scope and level of priority, and was

considered critical for understanding the impact genomic knowledge might have on individuals and society.

What will the next 50 years of medical science look like?

Having the essentially complete sequence of the human genome is similar to having all the pages of a manual needed to make the human body. The challenge to researchers and scientists now is to determine how to read all these pages, understand how the parts work together, and discover the genetic basis for health and the pathology of human disease. Genome-based research will eventually enable medical science to develop highly effective diagnostic tools, to better understand the health needs of people based on their individual genetic make-ups and to design new and highly effective treatments for disease.

When can we expect new and better drugs?

It usually takes more than a decade for a drug company to conduct the kinds of clinical studies needed to win marketing approval from the Food and Drug Administration (FDA) but some advances have already been made. Today, FDA requires pharmacogenomic labeling information for the labels of 106 medications. That means the label alerts doctors to genomic information that should be considered before prescribing that drug to a particular patient. Before the Human Genome Project, only four drugs carried such a label.

How has the Human Genome Project affected biological research?

HGP has given biological researchers a powerful tool to understand the genetic basis of health and illness. Moreover, it has had an impact on the way biological research is conducted. Traditionally, it has been a very individualistic enterprise, with researchers pursuing medical investigations independently. The magnitude of both the technological challenge and the financial investment prompted the Human Genome Project to assemble interdisciplinary teams, encompassing engineering and informatics as well as biology; automate procedures wherever possible; and concentrate research in major centers to maximize economies of scale. Such large, interdisciplinary teams have become common since the HGP's completion.

Now that the genome is complete, what's next for NHGRI?

NHGRI's vision for the future was published Feb. 10, 2011, in the journal *Nature.* The new plan envisions scientists identifying the genetic basis of most single-gene disorders and gaining new insights into multi-gene disorders in the next decade. This should lead to more accurate diagnoses, new drug targets and the development of practical treatments for patients currently without therapeutic options.

For more detailed information on NHGRI, the Human Genome Project and the future of genomics, go to:

- The NHGRI website: www.genome.gov
- The NHGRI Strategic Plan: Charting a course for genomic medicine from base pairs to bedside http://www.genome.gov/Pages/About/Planning/2011NHGRIStrategicPlan.pdf