

Lecture 39: Human Genome Project

The idea of the Human Genome Project first began in the 1970s when biologists started scrutinizing human gene at the molecular level. In 1980s several countries started to map parts of the human genome. In 1989, the Human Genome Organization (HUGO) was founded by eminent scientists of the field for promoting international collaboration for Human Genome Project related research. Systematic and collaborative research on Human Genome Project was started in 1990 with an aim to complete human genome sequence in 15 years. More than 2,000 scientists from over 20 institutes in six countries collaborated for this research. Finally, the first human genome draft was published in 2001 in *Nature* and *Science*, top ranked scientific journals.



The International Human Genome Sequencing Consortium published their results in *Nature*, 409 (6822): 860-921, 2001. "Initial Sequencing and Analysis of the Human Genome"

Celera Genomics published their results in *Science*, Vol 291(5507): 1304-1351, 2001. "The Sequence of the Human Genome"

In February 2001, the publicly funded Human Genome Project Organization and the private company Celera jointly declared that they had mapped the lion's share of the human genome. These maps show that there are only about 30,000 genes – much fewer than the 100,000 expected. In April 2003, the 50th anniversary of the publication of the structure of DNA, the completed map, was announced. The final sequence covers 99 per cent of the gene-containing regions of the genome. Human genome was completed by 2003. *Nature* published a special issue on human genome project in 2006. The human genome data opens new opportunities for research in medical biotechnology. All articles of this special issue are highly recommended for further reading.

<http://www.nature.com/nature/supplements/collections/humangenome/commentaries/>

<http://www.nature.com/nature/supplements/collections/humangenome/index.html>

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Human Genome Collection

It is now more than 15 years since work began sequencing the 2.85 billion nucleotides of the human genome. While the draft sequence was published in *Nature* in 2001, researchers at the Human Genome Project continued to fill the gaps and subject individual chromosomes to ever more detailed analyses. *Nature* is proud to present here the complete and comprehensive DNA sequence of the human genome as a freely available resource. Produced with support from our sponsors, *Nature* carries sole responsibility for all editorial content.

COMMENTARIES

New content to mark the publication of the Human Genome Collection are available here alongside links and original articles from the draft genome publication.

VIDEO

Enjoy exclusive interviews with scientists and researchers behind the Human Genome Project with Nature's online video streaming. Requires Flash.

CHROMOSOMES

The individual human chromosome sequences are presented here accompanied by specially selected papers, articles and letters.

REQUEST A COPY

Details of how to obtain your print copy and digital edition of the Human Genome Collection are available here.

Human Genomes
download a free digital edition here

Sponsors page

Supported by:

- Applied Biosystems
- Office of Science
- National Institutes of Health
- National Human Genome Research Institute

The human genome project was coordinated by U.S. Department of Energy and the National Institutes of Health. The human genome data and several other resources (including educational resources) are listed in Human Genome Project Information webpage. Snapshot of the webpage is given below:

http://www.ornl.gov/sci/techresources/Human_Genome/home.shtml

genomics.energy.gov

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Human Genome Project Information

- About the HGP
- Ethical, Legal, & Social Issues
- Medicine
- Education
- Gene Gateway
- Research Archive

Post-HGP Progress

SITE INDEX

Completed in 2003, the Human Genome Project (HGP) was a 13-year project coordinated by the U.S. Department of Energy and the National Institutes of Health. During the early years of the HGP, the Wellcome Trust (U.K.) became a major partner; additional contributions came from Japan, France, Germany, China, and others. See our [history](#) page for more information.

Project [goals](#) were to

- identify all the approximately 20,000-25,000 genes in human DNA,
- determine the sequences of the 3 billion chemical base pairs that make up human DNA,
- store this information in databases,
- improve tools for data analysis,
- transfer related technologies to the private sector, and
- address the ethical, legal, and social issues (ELSI) that may arise from the project.

Though the HGP is finished, analyses of the data will continue for many years. Follow this ongoing research on our [Milestones](#) page. An important feature of the HGP project was the federal government's long-standing dedication to the [transfer of technology to the private sector](#). By licensing technologies to private companies and awarding grants for innovative research, the project catalyzed the multibillion-dollar U.S. biotechnology industry and fostered the development of new medical applications.

OF INTEREST

- Battelle Report: \$3.8 Billion Investment in HGP Drove \$796 Billion in Economic Impact, Created Jobs, and Launched Genomic Revolution ([May 2011](#)). See Response From *Nature* ([May 11, 2011](#))
- Gene Patent Ruling Raises Questions for Industry ([Nov. 2, 2010](#), NYT)
- New Gene Tests Aimed at Reducing Colon Cancer ([Oct. 28, 2010](#), NYT)
- The Once and Future Genome ([June 25, 2010](#), Seed Magazine)
- Biology 2.0 Special Report on the Human Genome ([June 17, 2010](#), The Economist)
- The Genome at 10: Two-part article ([June 12](#) and [June 14, 2010](#), NYT)
- Human Genome at 10: 5 Breakthroughs, 5 Predictions ([Mar. 31, 2010](#), National Geographic)
- Disease Cause is Pinpointed with Gene ([Mar. 10, 2010](#), NYT)
- Cost of Decoding a Genome is Lowered ([Aug. 10, 2009](#), NYT)

What's next?

DOE GENOMIC SCIENCE
1998-2003
2003-2010
2010-2015
2015-2020
U.S. DEPARTMENT OF ENERGY

The screenshot shows the 'Exploring Our Molecular Selves' multimedia download page. It features a navigation menu on the left and a main content area with four interactive activity boxes. Each box includes a thumbnail image, a brief description, and download links for PC and Mac. The activities are: 'Milestones in Genetics: Timeline', 'Genes, Variation, and Human History', 'How to Sequence a Genome', and 'Ethical, Legal, and Social Implications (ELSI)'. A 'Back to Top' link is provided below each activity box.

The Human Genome Project
Exploring Our Molecular Selves

Welcome to the Human Genome Project, Exploring our Molecular Selves multimedia download page.

This page allows you to download all of the component parts that make up the award winning educational CD for The Human Genome Project.

Click on the PC or MAC DOWNLOAD text to download files.

[Genetics Timeline](#) [Genes, Variation, and Human History](#) [How to Sequence a Genome](#)
[Ethical, Legal, and Social Implications](#) [Glossary of Genetic Terms](#) [3D Video](#)

Milestones in Genetics: Timeline
An interactive timeline of milestones in genetics, presenting more than 90 key events and discoveries. By clicking on any year in the timeline, the user can read a short, illustrated story and often find archival images and original scientific publications.
[DOWNLOAD PC \(4.82MB\)](#)
[DOWNLOAD MAC \(3.7MB\)](#)

[Back to Top](#)

Genes, Variation, and Human History
Two interactive classroom activities that address what it means to be human – as a species, as a population, and as an individual. The user will learn how genetic variation allows the study of similarities and differences among individuals and how to use genetic data to compare human populations from various parts of the world.
[DOWNLOAD PC \(4.17MB\)](#)
[DOWNLOAD MAC \(2.9MB\)](#)

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How to Sequence a Genome
Animated and narrated segments presenting all the essential steps in sequencing a genome.
[DOWNLOAD PC \(11.82MB\)](#)
[DOWNLOAD MAC \(11MB\)](#)

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Ethical, Legal, and Social Implications (ELSI)
Seven case studies including background sections and information necessary to evaluate the scenario, short vignettes raising an important ethical or societal issue, a set of discussion questions, and web links and other resources to assist those in search of more detailed information.
[DOWNLOAD PC \(2.83MB\)](#)
[DOWNLOAD MAC \(2MB\)](#)
[VIDEO QT \(5.4MB\)](#) [Get PLUGIN](#)
[VIDEO WIN \(2.6MB\)](#) [Get PLUGIN](#)

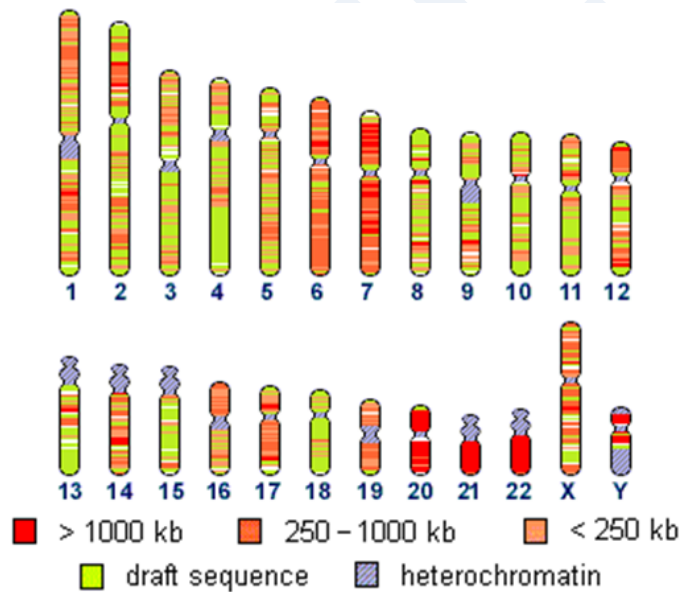
Students are recommended to view educational video about Human Genome project available on <http://www.genome.gov/Pages/EducationKit/download.html>

Human Genome Project also studied ethical, legal, and social issues (ELSI) surrounding availability of genetic information and 3% to 5% of total budget was used for the purpose.

Objectives of Human Genome Project

- Mapping and Sequencing the Genomes of Model Organisms.
- Data Collection and Distribution.
- Ethical, Legal, and Social Considerations.
- Research Training.
- Technology Development and Transfer.

Human Genome Sequencings published in 2001



	Total sequence (kb)	Non-redundant sequence (kb)	Percentage of genome
Finished	1,140,365	1,040,372	32.50%
Unfinished	3,547,899	1,951,344	61.00%
Total	4,688,264	2,991,716	93.50%

Source : NCBI

Additional sequencing was done after publication of human genome in 2001 to close gaps, reduce ambiguities. Single error every 10,000 bases in human genome sequencing ensures very high standard of the project. The first aim of human genome project was to determine

the location of each gene on 44 autosomes and 2 sex chromosomes and sequence each of the genes. The human genome consist of 4GB (one gigabase=10⁹bases). Amount of information coming out of human genome project was huge. It has been estimated that if all DNA base sequences of human genome project printed on small letters 220000 pages are required.

Issues arising from human genome project

1. This will enable us to ascertain whether fetuses are likely to develop any genetic disorder. Although the gene therapy is infancy and further research is required, but it is possible in future to replace bad genes by good genes. Several ethical issues are related with this.
2. Consider screening in uterus for diseases that do not get noticeable until many years after birth. Should we be screening for Huntington's disease or other diseases in uterus? Is it fair for the parents to make a decision about aborting a fetus because that fetus may have a crippling and develop fatal disease after 45 years of its birth? Do we want the child to acquire this potentially explosive information? How does it affect a person psychology to know that he or she will suffer from this cruel disease sometime later in life?
3. There are many potential liability issues that may arise out of the accessibility of genetic testing. Firstly, physicians who know of the existence of genetic screening and do not offer it to the patient may be legally responsible, just as they would be liable for failing to provide a non genetic diagnostic tool. Secondly, physicians could be accountable for revealing confidential genetic information or for not revealing it. A genetic screening may point out that the patient's siblings are at great risk of some genetic condition. Should they be informed of that risk even though that would be a violation of secrecy? Or if they do not tell the third party, is this a violation of responsibility?
4. Employers may seek to do a genetic screen of their employees to ensure that those that are prone to some risk do not suffer exposure. On the other hand, screens could be used by employer to appoint only persons improbable to be subject to any illness. Eventually pre-employment genetic screening would actually eliminate some people from the job market.
5. Some skills for example-mathematical ability are partly genetically based. Should genetic information be used to follow student's trade school versus university? How is the use of genetic information distinctive from the use of IQ? What is the proper genetic make-up of a doctor? Do we want more scientific brilliance or compassion? That is not a scientific question but a social policy question.
6. Genetic information could be beneficial in the criminal justice system beyond DNA fingerprinting. Some criminal defendants may have a genetic tendency to commend definite type of crime.

7. The health insurance system may ask for genetic information for an individual and may not cover a person with potentially costly genetic disorder.
8. Should government be permitted to maintain a bank of genetic information as many governments do now with fingerprints? This is very important information about each one of us.

Home assignments

1. The human genome project budget was approximately 3 billion US dollars. Do you think that this will be money well spent?
2. Would you like to know make-up of your own genome structure? Who else should know this information? Who should not have this information?