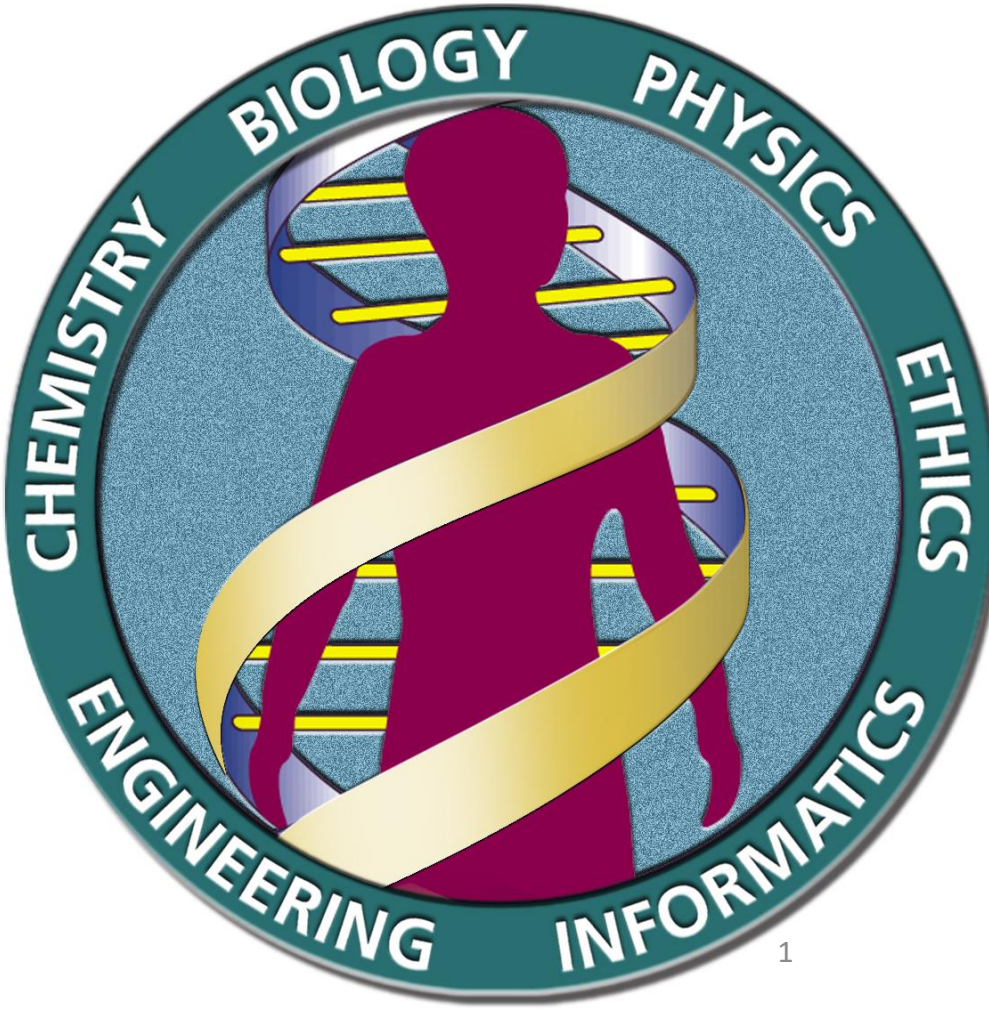
Human Genome Project

14 – 11 - 2022



Introduction

- The Human Genome Project (HGP) is an international, interdisciplinary, scientific research project aimed at determining the sequence of chemical base pairs which make up human DNA, mapping the entire human genome, and identifying its complex structures and functions.

- An early genome project was established in 1987 **(National Human Genome Research Institute – NHGRI 2012)** under the direction of the National Institutes of Health and the US Department of Energy, entailing a 15-year, $3 billion, plan to complete the human genome sequence.

President Bill Clinton and Francis Collins, M.D., Ph.D., (NHGRI Director) at a June 2000 event at the White House celebrating the draft human genome sequence generated by the Human Genome Project. Dr. Collins served as the de facto leader of the International Human Genome Sequencing Consortium, the group that sequenced the human genome during the Human Genome Project.

- Parallel to the US government sponsored HGP, the American researcher **Craig Venter, through his firm Celera Genomics**, announced in 1998 his intention to build a unique genome-sequencing facility to determine the sequence of the human genome over a 3-year period.

Aim and Objective

Human Genome Project

To Sequence the whole genome with 3billion bps

Creating a physical Map of the Human genome

To identify all the Disease causing genes

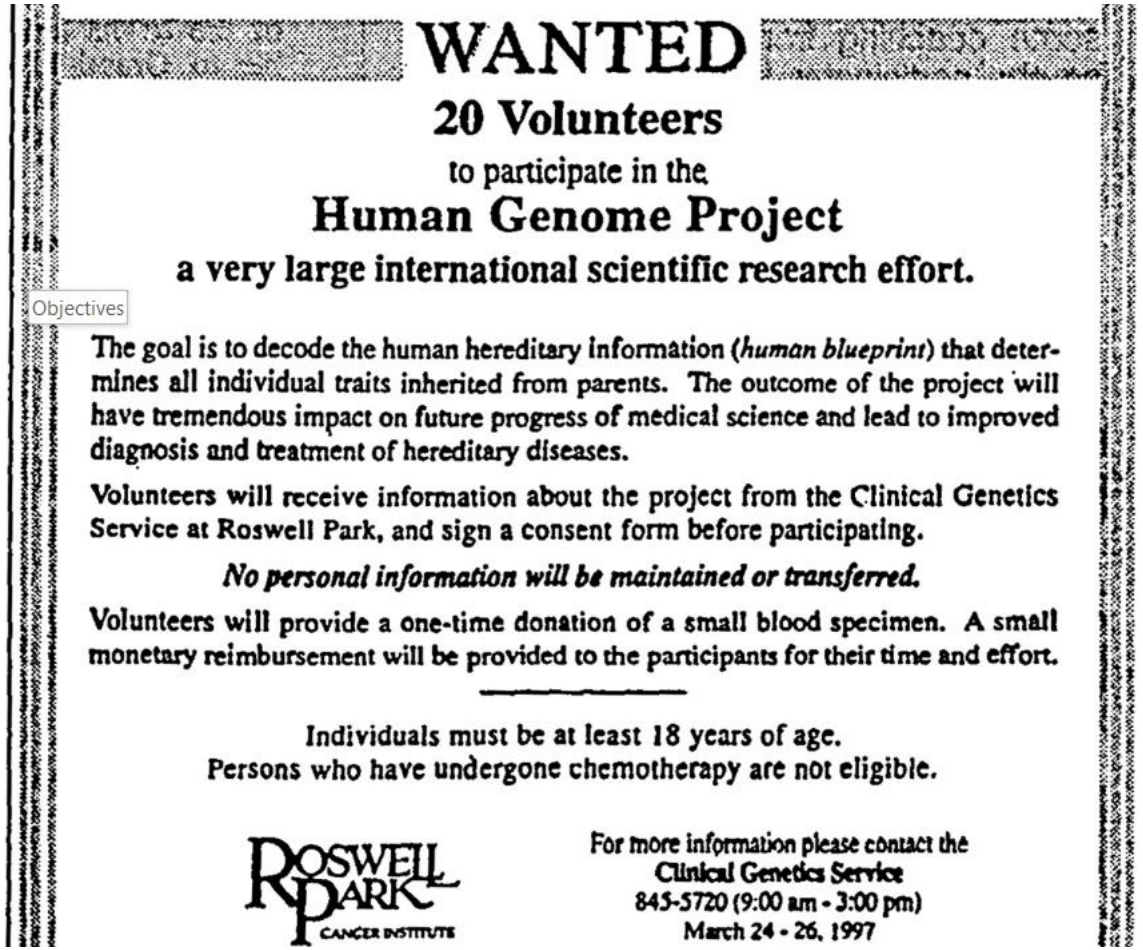
To understand the Function of Genes

To make the informations Available for all Researchers

To develop tools For processing and Analysing data

- After the humangenome sequencing was complete, the US Department of Justice filed a court brief stating that genes should not be eligible for patents because they are products of nature.
- Thus, the human genome database is publicly available to anyone (see The Genome Database – GDB, gdb.org).

Whose DNA was sequenced by the Human Genome Project? How was it collected?



WANTED

20 Volunteers

to participate in the

Human Genome Project

a very large international scientific research effort.

Objectives

The goal is to decode the human hereditary information (*human blueprint*) that determines all individual traits inherited from parents. The outcome of the project will have tremendous impact on future progress of medical science and lead to improved diagnosis and treatment of hereditary diseases.

Volunteers will receive information about the project from the Clinical Genetics Service at Roswell Park, and sign a consent form before participating.

No personal information will be maintained or transferred.

Volunteers will provide a one-time donation of a small blood specimen. A small monetary reimbursement will be provided to the participants for their time and effort.

Individuals must be at least 18 years of age.
Persons who have undergone chemotherapy are not eligible.

ROSWELL PARK CANCER INSTITUTE

For more information please contact the
Clinical Genetics Service
845-5720 (9:00 am - 3:00 pm)
March 24 - 26, 1997

The sequence of the human genome generated by the Human Genome Project was not from a single person.

It was a patchwork from multiple people whose identities were intentionally made anonymous to protect their privacy.

Most of the human genome sequence generated by the Human Genome Project came from blood donors in Buffalo, New York; specifically, 93% from 11 donors, and 70% from one donor.

Technologies used in HGP

- The Human Genome Project was aided by several 'breakthrough' technological developments,
  - Sanger DNA sequencing and its automation,
  - DNA-based genetic markers,
  - large-insert cloning systems
  - the polymerase chain reaction.
  - capillary-based sequencing
  - methods for genotyping single-nucleotide polymorphisms

Who carried out the Human Genome Project?

- international consortium of thousands of researchers .
- 20 separate universities and research centers across the United States, United Kingdom, France, Germany, Japan and China.
- The groups in these countries became known as the International Human Genome Sequencing Consortium.

Did the Human Genome Project produce a perfectly complete genome sequence?

- In June 2000, the International Human Genome Sequencing Consortium **announced** that it had produced a draft human genome sequence that accounted for 90% of the human genome. The draft sequence contained more than 150,000 areas where the DNA sequence was unknown because it could not be determined accurately (known as gaps).
- In April 2003, the consortium **announced** that it had generated an essentially complete human genome sequence, which was significantly improved from the draft sequence. Specifically, it accounted for 92% of the human genome and less than 400 gaps; it was also more accurate.
- On **March 31, 2022, the Telomere-to-Telomere (T2T)** consortium announced that had filled in the remaining gaps and produced the **first truly complete human genome sequence**.