Unit 2 - Genomics, Proteomics and Transcriptomics

Concepts of Genomics:

Introduction to Genome sequencing

- Human Genome project
- Genomic elements, SNPs, and genome-wide association studies.

Sequencing methods:

Sanger's dideoxy method,
NGS – Next Generation Sequencing – platforms and technologies

Whole Genome Sequencing

Exome sequencing

Introduction to Genome sequencing

- Genome sequencing is a method that determines the complete DNA sequence of an organism's genome.
- It can provide insights into the genetic basis of disease, evolutionary relationships between species, and the function of genes and non-coding regions of the genome.
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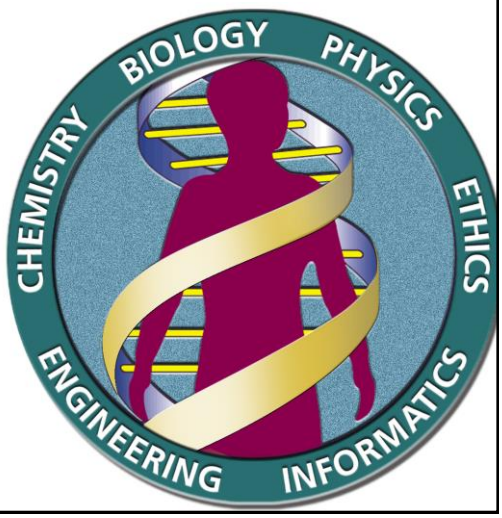
Human Genome Project

06-08-2024



Introduction

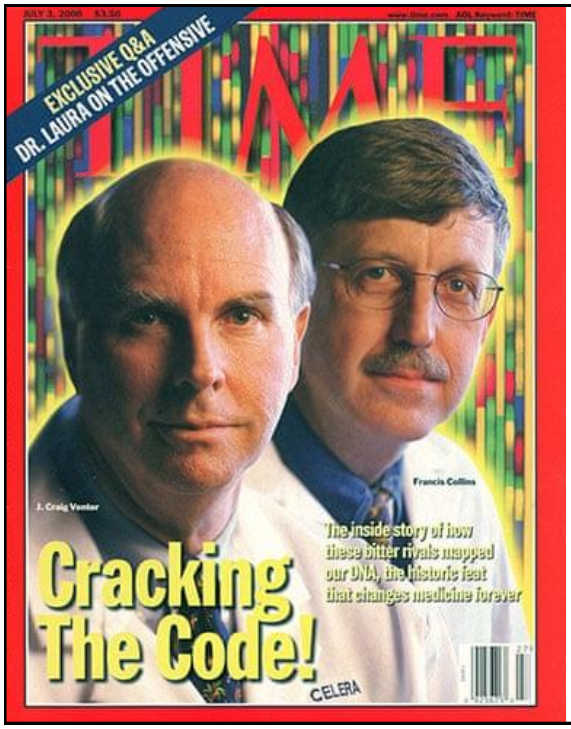
- The Human Genome Project (HGP) is an international, interdisciplinary, scientific research project aimed at determining the sequence of chemical base pairs which make up human DNA, mapping the entire human genome, and identifying its complex structures and functions.

- Reference : https://www.genome.gov/about-genomics/educational-resources/fact-sheets/human-genome-project

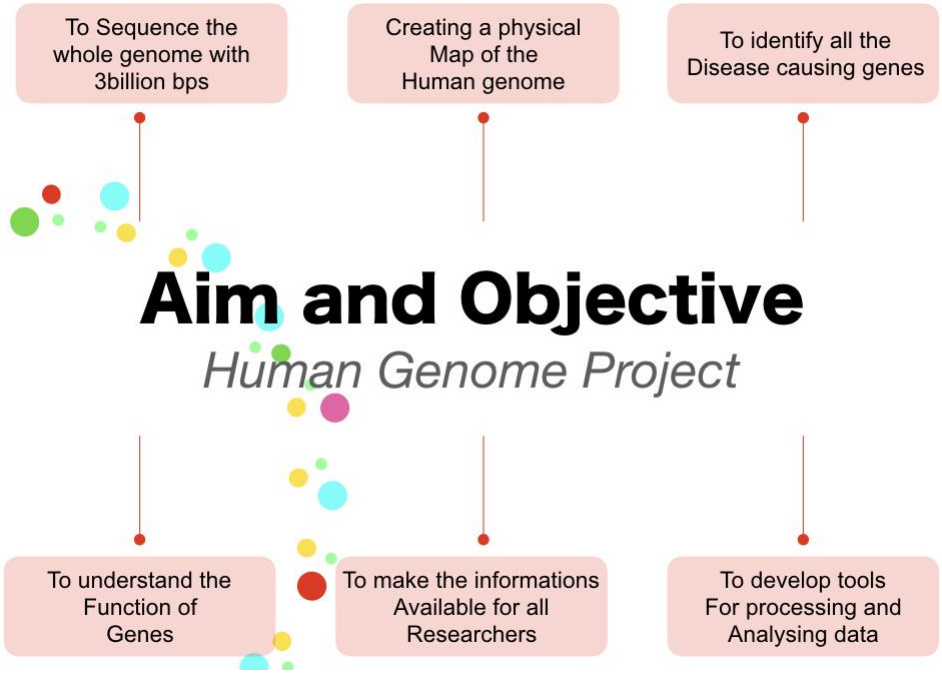
- An early genome project was established in 1987 **(National Human Genome Research Institute – NHGRI 2012)** under the direction of the National Institutes of Health and the US Department of Energy, entailing a 15-year, $3 billion, plan to complete the human genome sequence.

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.



- The **first analyses of the draft human genome sequence** were reported in the **February 2001 issues of Science and Nature**.



- After the human genome 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.

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 7% 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
  - NGS – Next Generation Sequencing

Who carried out the Human Genome Project?

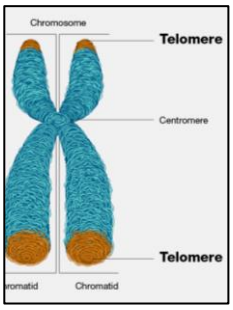
- international consortium of thousands of researchers .
- 20 separate universities and research centers across the United States, United Kingdom, France, Germany, Japan and China.
- The groups in these countries became known as the International Human Genome Sequencing Consortium.

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

- In June 2000, the International Human Genome Sequencing Consortium announced the a **draft human genome sequence** that accounted for 90% of the human genome. The draft contained gaps (150,000) that could not be determined accurately.

- In April 2003, the consortium **announced** the 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**.



Applications

1. It helps in the **identification of mutations** linked to different forms of **cancer**.

2. Gene discovery aids in developing **gene-based treatments** for hereditary and acquired diseases.

3. The human genome project will help to **assess risks** posed to individuals by **environmental exposure to toxic agents.**

4. Detailed genome maps have aided researchers in identifying **genes associated with genetic conditions**, including fragile X syndrome, inherited colon cancer, Alzheimer's disease, and familial breast cancer.

5. **DNA Forensics**: Identify potential suspects whose DNA may match evidence left at crime scenes.

6. The Human Genome Project has provided a wealth of new opportunities for researchers to **explore the genetic basis of human disease** and to develop **new approaches to diagnosis and treatment.**