

## The Spiraling Effect Created by The Human Genome Project

Rebecca Tochilnikov

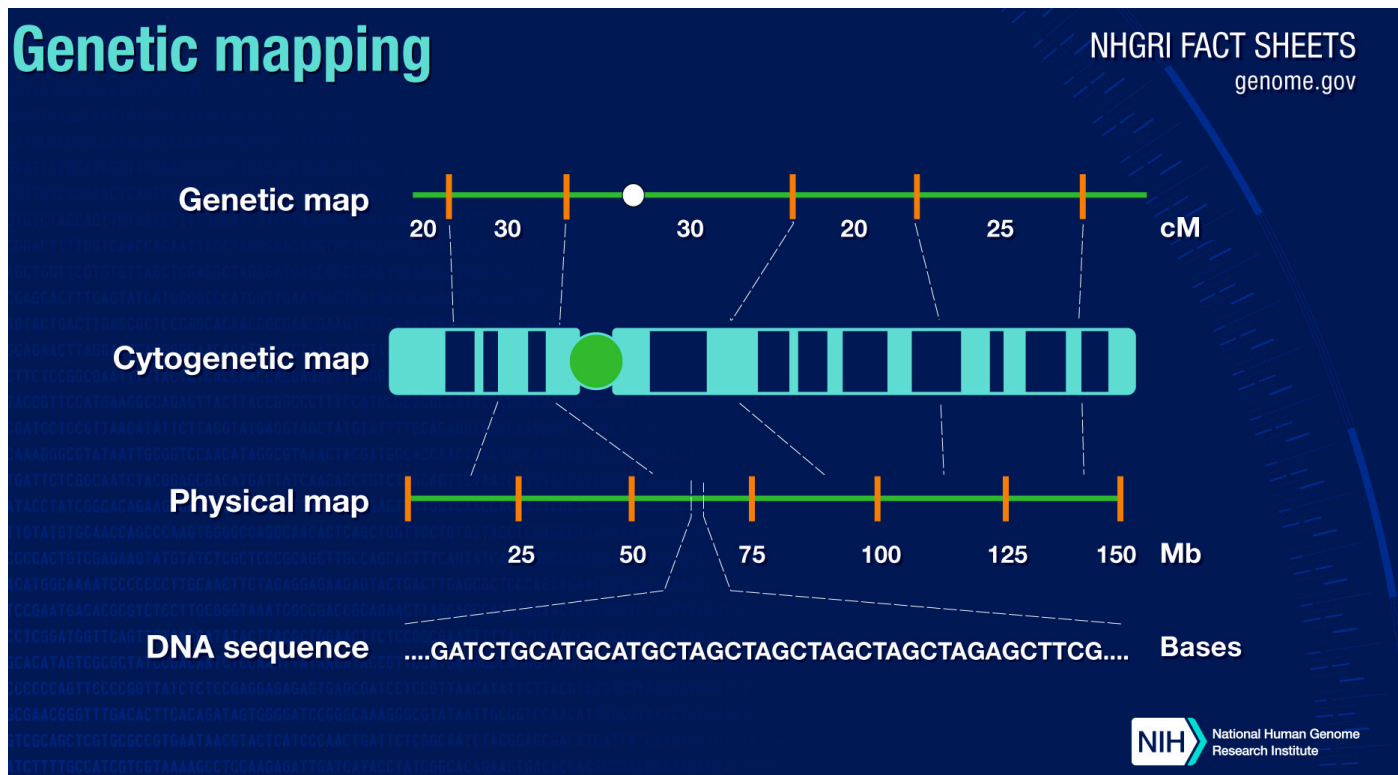
### I. Abstract:

The Human Genome Project did not happen in isolation – breakthroughs lead to debates and examinations of ethics. Mapping out the entire human genome was a revolutionary achievement that changed medicine and scientific thinking forever. This research also examines the reactions, especially the ethical concerns; and how those led to reforms in medical research and privacy laws. The literature review began with official press releases announcing the project's completion and speeches from key figures like James Watson, who contributed to diagnosing the molecular structure of DNA. Many scientific articles and documentaries, like PBS's *Cracking the Code of Life*, provided strong context of the project's impact on society and medicine. Further research covered the ethical issues raised, such as fears of genetic discrimination and “designer babies”. This research is organized around the three aspects of the project's innovations: *Revolution, Reaction, Reform*. Revolution refers to the breakthrough mapping of the human genome; reactions to how the scientific community and the public ranged from amazement to ethical dilemmas; reforms to how we continue to transform medical research and legal protections. The Human Genome Project was more than just a scientific milestone; it sparked important reactions and changed how humans think about genetics and privacy, leading to lasting reforms in medicine and ethics.

### II. Introduction:

Former Director of National Human Genome Research Institute (NHGRI) Francis S. Collins once declared, “I think there are people whose lives have been saved because of the study of the genome.” The Human Genome Project goal was to sequence the entire genome, which allowed scientists to understand the reality of human functioning by analyzing cells and genetics through special technology in advanced labs; the project succeeded in mapping tens of thousands of human genes. America was not the first country to build on this idea; it started with three separate endeavors competing against each other and then combining together into this project. The project's development started in 1990 and ended in 2003, in total, its completion took thirteen years, costing about three billion dollars. Many scientists believe the cost was worth it, since the project improved medical professionals' understanding of the fundamentals of human DNA and life in general. With these advancements, they were able to create a way to prevent inherited diseases from forming through gene therapy. However, The Human Genome Project caused multiple concerns and uncertainties ethically, as many were nervous that scientists would misuse genomic information when it comes to insurance and employment. Additionally, people had religious concerns about gene editing and designer babies, arguing that it was not part of “God's plan”. The Human Genome Project represented a revolution in modern medicine by shifting genetics from abstract theory to a complete map of the human genome between 1990 and 2003. Although it provoked strong reactions over privacy, discrimination, and religious viewpoints, yet on the other hand in turn drove reforms such as new bioethical standards, gene-therapy practices, targeted cancer treatments, and laws to protect genetic information. Through this, The Human Genome Project fundamentally reshaped how scientists, lawmakers, and physicians understand and use human genetic information, to improve patient care and protect genetic privacy.

**Figure 1. Genetic Mapping** – The diagram shows how scientists created different types of genetic maps while studying the human genome. It shows the different types of chromosome maps used to precisely mark the DNA sequences (made out of individual base pairs). The Cytogenetic Map illustrates the structure of a chromosome and shows how specific locations on the chromosomes, called a loci, contain genes that code for particular human traits. The sections above and below the cytogenetic map represent calculated distances between genes based on how often they cross over during meiosis. This process was essential to the project due to the sequencing process that gave scientists a complete picture of the human genome.



### III. Revolution

During the Scientific Revolution, there was a drastic change in the perception of science: it became thought of as a “A new view of nature,” where science also modernized into using more technological methods and using more quantitative views instead of qualitative views (Brush and Spencer). The Human Genome Project shifted the perception of the importance of human DNA and genetics, just like the Scientific Revolution shifted to the scientific method. The Human Genome Project changed science and medicine internationally, by providing a new foundation for understanding human existence through analyzing the DNA. This revolutionary project led scientists to discover “about 99 percent of the human genome's gene-containing regions,” and it was “sequenced to an accuracy of 99.99 percent” (NIH). DNA are complex instructions that map out specific sequences that have certain functions in each person's body. That 99% that has been identified makes it easier for doctors to identify issues in people's genes where they can possibly edit and change the person's life by making them live longer in a healthier way. That level of understanding represents a revolutionary shift in medicine, because it moves scientific practice from observation to precise intervention. As scientists began

decoding the undiscovered complexity of human DNA, they recognized the genomes' potential to guide future discoveries, noting that "Unravelling the human genome first and foremost provided us with a road map for the study of human genetics, helping us to discover new genes and new gene functions" (Potters). By mapping the human genome, scientists created a new international foundation for future research that fundamentally changed studying techniques, diagnosis methods, and treating gene diseases.

This roadmap allowed scientists to identify specific genes and understand their functions, which transformed medical research from hypothesising into precision. It also informed the development of gene therapies and personalized medication, showing the revolutionizing approach to changing mental health and disease.

The project was achieved uniquely at an international scale through their collaboration together instead of against each other, in order to create a revolutionary accomplishment that will forever change the perspective of science in the medical industry. Specifically, "The decoding of the genome turned out to be an intriguing competition (and in the final stages, collaboration) between a consortium of world-wide government-paid scientists on the one hand and the private company Celera" (Potters). That collaboration demonstrates how The Human Genome Project revolutionized scientific knowledge into scientific practice and interaction, showing that global cooperation can achieve more than individual countries. This cooperation allowed for faster progress, higher accuracy, and broader sharing of intellect, highlighting knowledge and research is done best when shared.

#### IV. Reaction

Every human on Earth has cells, and if they have cells, they have DNA that has been sequenced by scientists. Each individual has their own variation, for which The Human Genome Project created the foundation for further analysis. The sequencing of the human genome caused a wide range of reactions worldwide, from excitement about medical possibilities to concern over ethical dilemmas. Many people saw the project as a beacon of hope, especially for the prevention and treatment of genetic diseases. As one Bioethics source notes, "Early diagnosis of a genetic disease may facilitate the adoption of treatments for the total or partial correction of the disease or any of its manifestations" (Bioethics Observatory). The Human Genome Project created optimism and hope, as people envisioned a future where genetic diseases could be detected early on and prevented. This excitement caused a reaction of more scientific creations like CRISPR; The Human Genome Project was known for the mapping of human DNA, so through that scientists devised that Clustered Regularly Interspaced Short Palindromic Repeats (CRISPR), which can correct or treat genetic disorders, transforming medical care and offering new opportunities for healthier lives worldwide.

CRISPR is one of the most progressive tools to emerge from genomic research, which "is a natural defense mechanism found in bacteria that helps them fight off viruses," researcher, "Jennifer Doudna and Emmanuelle Charpentier discovered a DNA-cutting protein, called Cas9, in the CRISPR system," then continued experimenting so "they could control Cas9 using 'guide RNA' that directs the protein to cut particular locations in the genome, giving them the ability to cut any DNA molecule with precision" (NSF, *CRISPR: A Biotech Breakthrough*). As an outcome of the genome mapping, CRISPR represents a groundbreaking reaction from the knowledge

previously gained. By turning the bacterial defense system into a precise tool for editing DNA, scientists like Doudna and Charpentier applied their knowledge of the genome into practice. CRISPR allows scientists to target specific genes and precisely cut or modify DNA sequences, enabling the advancement of biomedical research. Scientists use two main components – the Cas9 protein and a guide RNA; the guide RNA leads the Cas9 to a specific sequence in the DNA, after Cas9 acts like molecular scissors that cut the DNA into that spot, then the cell's natural repair mechanism will add or remove DNA.

The creation of The Human Genome Project led to gene editing, CRISPR, and personalized medicine; global dilemmas and profound ethical questions were raised about how far humans should intervene in the natural selection of genetic processes. According to the Bioethics Observatory, “we can be skeptical that this is the case in controversial areas such as the application of genetic diagnosis for eugenic purposes, as is the case with preimplantation genetic diagnosis for the selection and/or elimination of embryos, or prenatal diagnosis in order to find out if the fetus has potential alterations due to genetic defects in its DNA or chromosomal makeup” (Bioethics Observatory). Gene editing and personalized medicine offer hope for preventing disease, they also raise questions about eugenics, embryo selection, and the moral limits of human intervention. Many people with a religious perspective are questioning gene-editing: Christians, Jewish, Muslim, and Buddhist.

The Christian perspective emphasizes careful engagement with science. In her article “GenEthics and Religion,” bioethicist Martina C. Corne explains that “many theologians, however, show a distinctive interest in natural sciences, such as the Augustinian monk Gregor Mendel, the founder of modern genetics. A scientist’s daily work involves a profound and close study of creation which permits him a very direct insight into its ‘wonders’ and helps him develop great respect for its power” (Cornel). Christian values respect scientific inquiry, highlighting that ethical considerations should guide how humans use genetic technologies like gene editing.

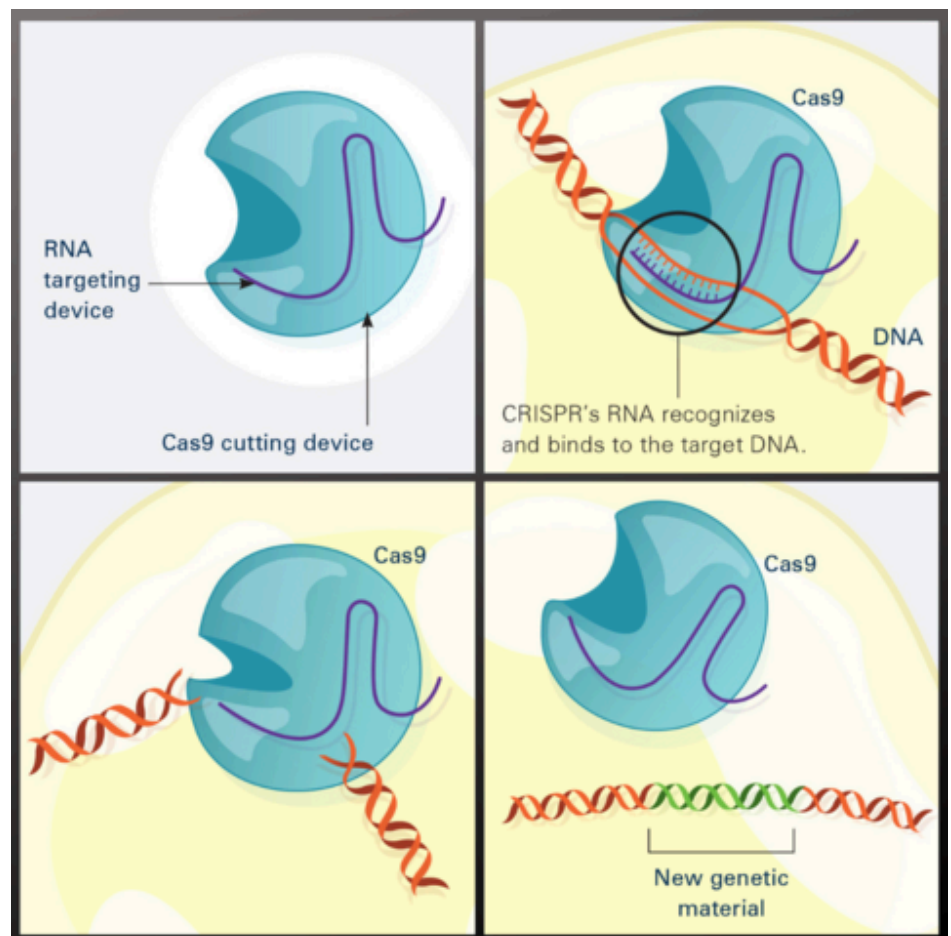
The Muslim community emphasized caution in genetic interventions, noting that “On the beginning of human life, there is a general consensus that there is potential life in early embryos and they must be treated with caution. The intention to eliminate diseases may be justified in actions that may bring about the possibility of embryo destruction. This sometimes is interpreted to be the lesser of two evils” (Cornel). Islamic ethics balance the potential benefits of genetic interventions with moral responsibility to protect early human life, emphasizing careful deliberation in gene editing practices, while prioritizing adherence to God's guidance.

Juxtaposing, the Jewish belief emphasizes that “At its roots, Judaism is a tradition that sees human beings as ‘co-creators’ with God in creation and that does not exhibit a fear that human beings will use technology to ‘play God’” (Cornel). In this viewpoint, Judaism generally supports the ethical use of genetic technology, viewing humans as partners with God in improving life rather than being restricted from intervening in natural processes.

Similarly, the Buddhist perspective highlights the dignity and rights of embryos, suggesting that “A less liberal attitude applies to the beginning of life. An embryo is human and thus possesses human dignity and human rights at the time of conception. In Buddhism, persons are interdependent. Germline cell therapy for instance is ethically questionable due to

its potentially negative effects on humanity” (Cornel). Buddhist culture prioritizes the protection of human life and considers the broader ethical impact on humanity, making germline gene editing ethically questionable. Overall, Christian and Muslim interpretations share similar values that human genes are sacred and the gene-editing is seen as unethical, while Judaism and Buddhism focus more on ethical responsibility and reducing suffering rather than forbidding edits because they are not “god-given”.

Beyond religious aspects, ethical discussion around gene editing also emphasizes the protection of individuals and the responsible use of genetic information. As the Bioethics Observers notes, “It is considered that, although the human genome is regarded as the ‘heritage of humanity’, individual persons must be protected and the intended use of the information in the applied field must be taken into account” (Bioethics Observatory). This highlights that regardless of cultural or religious beliefs, society must carefully regulate how genomic data and technologies like CRISPR are applied to prevent misuse, discrimination, or exploitation. Ethical safeguards are therefore essential to ensure that scientific advancements in personalized medicine and gene editing benefit humanity and individual rights.



**Figure 2. CRISPR-CAS9 Gene Editing** – This model shows how CRISPR-Cas9 gene editing works to change DNA. Step 1: the guide RNA attached to the Cas9 enzyme. The RNA guides the Cas9 protein to a specific sequence in the DNA strand. Step 2: the guide RNA recognizes and then binds to the matching DNA sequence in the genome. This ensures the enzyme edits in the right location. Step 3: The Cas9 enzyme cuts the DNA strand at that location. Step 4: After the completed cut, scientists can change or insert DNA, allowing them to replace the damaged genes and eliminate the genetic disorder.

## V. Reform

In the Reform stage, the National Human Genome Research Institute (NHGRI) highlights the long-term impact of genomic discovery, noting that “today, more than 1,400 disease genes have been identified” (NIH). The sheer number of gene discoveries outweigh the controversies of its applications. The new knowledge of identifying disease genes has changed political and social standards, which is an important aspect of protecting medical choices and people's rights.

Many genetic disorders, such as Cystic Fibrosis, Huntington's disease, and Tay-Sachs disease, had been without treatment for decades until the mapping of the genome (“Cracking the Code of Life”). The discovery of DNA sequences of specific genes being involved in hereditary disorders allows gene therapy techniques to be used: “This refers to the restoration of the defective gene in an individual with an inherited disease, either by inserting a functional gene into the genome, or by silencing, modifying the expression level, or editing the gene sequences of the altered gene” (Bioethics Observatory). Modifying a person's gene means that the malfunctioning gene would be muted, so that the disease symptoms have less impact on the person's life habits. Editing the gene sequence of the mutated gene revises it to operate precisely for a proper human health. Before the human genome was mapped, these therapies were not a possibility due to limited information and technology to test for genetic disorders, diagnose diseases, and treat accordingly.

Personalized medicine was developed rapidly after the Genome Project was completed, allowing many medical professionals to quickly help their patients choose treatments that are precise and effective for them. Recently, 2024 research from Széchenyi István University described how “Pharmacogenomics and genomics-driven treatments will enable personalized drug selection and dosing before starting a regimen, optimizing therapeutic outcomes” (Sustainable and Equal Integration of Genomics Into Healthcare). Pharmacogenomics studies the most appropriate pharmacological treatments for the individual genomic profiles of patients with genetic diseases. Previously, medicine was created to match certain characteristics of a disease for an average patient, but now in modern society every person's gene variants can be sequenced and analyzed, creating an opportunity for more personalized medical experiences in order to receive the adequate regimen. According to the US Food and Drug Administration (FDA), “[p]harmacogenomics can play an important role in identifying responders and non-responders to medication, avoiding adverse events, and optimizing drug dose” (Bioethics Observatory). In the past, those with genetic disorders were untreatable because their DNA was undocumented until the project's success of mapping it out. In order to analyze genetic profiles, new methods, including testing and AI, will start being more prominent in daily routines: “Genetic tests will become more accurate, results will be available sooner, and interpretation will be simplified through the utilization of artificial intelligence” (Sustainable and Equal Integration of Genomics Into Healthcare). Pharmacogenetics improves healthcare by allowing doctors to tailor treatments to a person's genetic makeup, rather than using the one-size-fits-all approach. This leads to safer and more effective medicine by reducing the harmful side effects and improving how well drugs work for individual patients.

The Human Genome Project has changed cancer research by allowing scientists to analyze genetic mutations in tumors, develop targeted drugs more efficiently, and create personalized treatments that improve outcomes for patients. Without the Human Genome

Project, new discoveries and technologies would have developed painstakingly over time. According to professionals at the Institute of Cancer Research, “The Human Genome Project initiative and technologies were the engine that started up the international Cancer Genome projects enabling the mapping of the genomes of hundreds of thousands of cancer patients. Through this, we were able to rapidly uncover cancer driver genes, and to discover drugs for those, at unprecedented speeds” (How the HGP Shook The World of Cancer Research). Its exigence is paramount to future medicine and society because the threats of widespread cancer taking over millions of people's lives needs to be controlled. Rapidly diagnosing and treating cancer during its early stages leads to better outcomes and an increase in survival rates. New drugs are created to better treat cancer based on scientists understanding and analyzing the results of the Human Genome Project. Specifically, “By being able to apply genomic technologies such as gene sequencing to drugs that are being developed, scientists can speed up the process by

figuring out in a more efficient way whether certain drugs act on their target, while also gaining insights into drug metabolism” (How the HGP Shook The World of Cancer Research). Cancer's urgency is always of the highest concern because this fatal disease must be addressed to allow someone to outlive it. The new drugs being created are more powerful due to the drug being able to detect the right cells – tumor cells – and target them with expedience. As an example, in 2021 “The Breast Cancer Association Consortium (BCAC) tested 60,466 women with breast cancer and 53,461 control individuals for a panel of 34 known or suspected breast cancer susceptibility genes. Of those 34 genes, only nine were confirmed as

	HGP Begins 1990	HGP Ends 2003	10 Years after HGP 2013
<b>Genome Sequencing</b>			
Cost to Generate a Human Genome Sequence	~\$1 billion	~\$10-50 million	~\$3-5 thousand
Time to Generate a Human Genome Sequence	~6-8 years	~3-4 months	~1-2 days
Human Genome Sequences	0	1	Thousands
<b>Genome Sequence Data</b>			
Total DNA Bases in GenBank	~49 million	~31 terabases	~150 terabases
Whole-Genome Shotgun Bases in GenBank	0	~9.6 terabases	~391 terabases
Vertebrate Genome Sequences	0	3	112
Non-Vertebrate, Eukaryotic Genome Sequence	0	14	455
Prokaryotic Genome Sequences	0	167	8760
Human Single-Nucleotide Polymorphisms	~4.4 thousand	~3.4 million	~53.6 million
<b>Human Genetics</b>			
No. Genes with Known Phenotype/ Disease-Causing Mutation	53	1474	2972
No. Phenotypes/Disorders with Known Molecular Basis	61	2264	4847
No. Published Genome-Wide Association Studies (GWAS)	0	0	1542
Replicated Disease-Associated Genetic Variants	0	6	~2900
<b>Genomic Medicine</b>			
Drugs with Pharmacogenomics Information on Label	4	46	106
<small>Since the beginning of the Human Genome Project 23 years ago, genomic data have steadily accumulated, laying the foundation for advances in human health. Data compiled from various sources by National Human Genome Research Institute.</small>			

high confidence breast cancer risk genes: *BRCA1*, *BRCA2*, *PALB2*, *ATM*, *CHEK2*, *TP53*, *BARD1*, *RAD51C*, and *RAD51D*" (Manolio, et al). Scientists were able to locate the specific genes and their cancerous risk due to the genome mapping which facilitated the cascading of several technologies. The discovery that there are only nine genes impacting breast cancer allowed their focus of developing drugs for treatment that can successfully remit cancer. According to the World Health Organization, "Cancer is a leading cause of death worldwide," so the significance of these new drug treatments is that more people will survive and be able to live a long prosperous life.

Another aspect of the societal reform yielded from The Human Genome Project are laws that defend individuals from discrimination based on their inherited DNA: Genetic Information Nondiscrimination Act (GINA). The law is used to protect people from unethical costs of medical insurance and unfair hiring practices based on genetic makeup: "GINA is a U.S. federal law that protects against genetic discrimination in the workplace and through one's health insurance. It also safeguards individual and family privacy of genetic information" (American Society of Human Genetics). The Human Genome Project expanded on personal DNA profiling which caused the legislation of ethical laws where people's unprecedented vulnerability is protected from unfair cost and treatment.

**Figure 3. Quantitative Advances Since the Human Genome Project (HGP)** – This chart highlights how the cost and time required to sequence a human genome decreased dramatically while the amount of genetic data and known gen-disease connections rapidly increased. Also, it shows how technological advancements allowed scientists to sequence thousands of genomes much faster than during the early years of the project. Overall, the model demonstrates how the projects accelerated genetic research and helped advance modern medicine and personalized treatments.

## VI. Conclusion

Ultimately, The Human Genome Project allowed scientists to gain knowledge on the function of DNA in the human body, manage gene defects through gene editing, and learn how to more accurately diagnose people with genetic diseases. Additionally, the project transformed cancer research by helping scientists identify genetic mutations linked to tumors, leading to the development of targeted therapies and more personalized cancer treatments that improve survival rates and patient outcomes. Despite all of its benefits, it generated ethical controversy within religious doctrine, resulting in people unwilling to participate in new life changing medical treatment. The project created precariousness in protecting one's privacy based on personal medical records; federal laws must ensure that employment and health insurance is not threatened. Revolution, reaction, reform: these represent the phases of The Human Genome Project, including revolution in the way biology was viewed, reaction in the ethical dilemmas, and reform in the legal and medical protections.

The hope of The Human Genome Project continues to be amplified with new applications like continuing to promote more advanced gene editing technologies, including those that use artificial intelligence to improve accuracy and patient care. These ongoing innovations built on the foundation of the The Human Genome Project, which has led to such expansion of ideas and medical discoveries, and thus will not only facilitate biological research, but also continue to



improve health for people despite their genetic challenges. In the years ahead there is a strong potential for genetic testing to become more accessible and affordable to the public, alongside continued growth in cancer research and genetic trials within hospitals. Expanding this work to address other current 'incurable' diseases such as diabetes and Alzheimer's, could open new pathways for treatment and prevention.



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