Extended Reading for Weekend March 25th, 2023

Enjoy, and have a nice day!

The Human Genome Project

1 In 1990, the United States National Institutes of Health (NIH) and the Department of Energy, in collaboration with partners in 18 countries, ***embarked*** on the Human Genome Project (HGP), the most ambitious venture **undertaken** since the Manhattan Project to develop the atom bomb or the Apollo project to put a man on the moon. At an **estimated** cost of $3 billion to complete the task by 2005, leading scientists and researchers in the field of molecular biology set out to identify all 30,000 to 40,000 genes belonging to the human genome and to map the location of the three billion base pairs of DNA—in other words, to write the Book of Life. This **definitive** resource was intended to lead to the understanding of genetic diseases, the creation of effective pharmaceuticals and medical treatments, and the ***alleviation*** and prevention of human suffering due to genetically **transmitted** diseases. In order to serve all of humankind and prevent control by any scientific, corporate, or national interest, all information was to be stored in public electronic databases and made freely and readily accessible to anyone who required it.

Historical background

2 Throughout history, people have turned to mythology, folklore, and religion for explanations of life's origins, and to this day there are many who firmly believe what has been written in religious books and passed down from generation to generation. With the advent of the Age of Reason in the eighteenth century and scientific advances in the nineteenth century, however, the quest for deeper knowledge could be satisfied by digging for **empirical evidence** and putting it to empirical test.

3 In 1865, Gregor Mendel, an Austrian monk who had been experimenting for eight years with garden peas, announced to the scientific community that specific characteristics, or traits, were transmitted from parent to offspring in an organized and predictable manner. Along with Charles Darwin's theory of natural selection and evolution as stated in his 1859 publication, *On the Origin of Species,* Mendel's work set the stage for the science of genetics to become the ***preeminent*** explanation of where life comes from. With the help of improved microscopes, scientists discovered the existence and structure of cells containing chromosomes. In the early 1900s, experiments with fruit flies **revealed** that chromosomes located in the cell's nucleus were made up of genes. The *Drosophila,* commonly called the fruit fly, was the first living organism to be genetically mapped. In 1944, Oswald Avery identified genes in bacteria as genetic messengers made of deoxyribonucleic acid (DNA). In 1953, James Watson and Francis Crick discovered the double helix structure of DNA, for which they received the Nobel Prize nine years later.

4 With each groundbreaking discovery, molecular biologists were able to form an ever clearer picture of the mechanics of life. To crack the code of life, prominent scientists proposed compiling a comprehensive genetic map of a human being. Simpler organisms, such as the fruit fly and

bacteria, had already been genetically mapped, but due to technical limitations, attempts with human genes had produced crude versions lacking precise detail. To duplicate, analyze, and store human DNA on the scale that was being proposed, more ***sophisticated*** tools and advanced technology would have to be developed.

5 In the 1980s, technology was making great strides. The development of recombinant DNA technology enabled researchers to split long strands of DNA into fragments and to splice and copy specific genes for study. Rapid advances in the 1960s and 1970s had produced machines like the polymerase chain reaction (PCR) machine, or DNA amplifier, that could duplicate DNA faster and cheaper. Developments in computer technology, in particular the invention of the silicon semiconductor chip, had made it possible for huge amounts of data to be analyzed at greater speeds and stored on more compact, portable, and affordable personal computers. Finally, the Internet provided a means for institutions to share and distribute information quickly and widely.

6 At the same time, the world was becoming more genomic. The discovery of the gene that led to Huntington's disease and the Federal Drug Administration's approval of synthetic insulin, biotechnology's first pharmaceutical product, ignited hopes that once the genetic causes could be determined of such debilitating diseases as muscular dystrophy, cystic fibrosis, and sickle-cell anemia, effective drugs and treatments for cancer and heart disease would eventually follow. **Emerging** biotechnology companies were making headlines, and their stocks were soaring on Wall Street. In general, biologists agreed that the project could be ***accomplished***, but not everyone believed that it should be done.

7 Despite **ethical** considerations and doubts that the project would bring about the desired results, **widespread** enthusiasm for the project's immense **potential** led to a series of meetings and conferences in 1986 and 1987 to set goals, to estimate the required outlay in money, time, and human resources, and to generate information for the government agencies and institutions that would provide the financial resources. When the U.S. Congress **allocated** funding to the NIH and the Department of Energy, the Human Genome Project was on its feet.

8 In September 1999, the Human Genome Project announced that 200 scientists working on three continents had assembled 25 percent of the entire genetic sequence. By February 2001, the HGP had published its first draft of 90 percent of the human genome in special issues of *Science* and *Nature*. In April 2003, two years ahead of schedule, the project succeeded in completing the sequencing of 99 percent of human genes to 99.99 percent accuracy, with 341 gaps. Even before its completion, the Human Genome Project and the information disseminating from it were opening doors in the fields of medicine, energy, the environment, agriculture, bioarchaeology, anthropology, and forensics.

Medical benefits

9 The Human Genome Project's ultimate goal was to provide **fundamental** genetic information that would lead to the treatment, and eventually the eradication, of many of the 4,000 genetic diseases and defects that afflict humans. As diagnostic genetic tests become more sophisticated and available, doctors will eventually put together genetic profiles for patients, determine their risk for disease, and make diagnoses before individuals become sick—or before they are even born. With the focus on preventing disease, doctors can begin to provide genetic counseling to families who want to understand their genetic background, as well as to couples who are planning a family. Advances in computer hardware and software will allow doctors to analyze biological samples more quickly and cost-effectively and to transfer the information to patients' computerized files, which they will then carry with them on computer chips. On the basis of this information, it will be possible to predict an individual's ***susceptibility*** to drugs and to environmental factors that are responsible for allergies.

10 The HGP will **revolutionize** not only how doctors treat patients but also how medical therapy is delivered, particularly in the emerging field of pharmacogenomics. An online article published on the Human Genome Project Information website predicts that up to 3,000 new drugs will have been developed, tested, and marketed by 2020. These products will generate sales in the billions of dollars for biotech and pharmaceutical companies, as researchers use genome targets to design and customize more effective drugs with fewer side effects, to **eliminate** adverse drug reactions in patients, and to make intervention more precise and successful. In addition, inexpensive vaccines will be ***engineered*** to activate the immune system without causing infections. In the long run, these improvements are expected to reduce the overall cost of health care. Although areas such as the cloning of organs for transplants have yet to be fully explored, the potential for life-saving and life-**enhancing** advances is vast and exciting.

Ethical, legal, and social issues

11 Although the Human Genome Project's picture of human health in the future appears rosy and immensely hopeful, it has its darker side. From the outset, the HGP **specified** as one of its goals the need to examine the ethical, legal, and social ***issues*** (ELSI) involved in making genetic information available. Between three and five percent of HGP's annual budget was allocated for this purpose. If the HGP is to serve humankind as intended, laws and regulations must prevent ***abuse*** and misuse of this information.

12 At the forefront of ELSI was the concern that employers and insurers could **discriminate** against employees and **deny** coverage on the basis of genetic test results. In May 2008, the Genetic Information Nondiscrimination Act (GINA) made it illegal for employers, insurers, courts, schools, and other entities in the United States to discriminate on the basis of genetic information. The potential for social ***stigmatization*** of individuals on the basis of their genetic makeup and the ensuing psychological suffering cannot be overlooked or minimized.

13 Sensitive issues of privacy, confidentiality, and ownership of genetic information that can only be dealt with through strict legislation are accompanied by philosophical and ethical issues. Scientists now know the location of genes on a chromosome, but it will take further research before they understand how genes work and how environmental factors come into play. This complexity makes it extremely difficult, if not impossible, for anyone to predict the outcome of what critics call tampering with nature or playing God. Although the idea of creating designer babies may seem attractive to some, the birth of a genetic elite brings to mind the practice of eugenics and the disastrous attempts of past regimes to create a superior race. Without clear ethical guidance, humankind's progress could end up in territory we should never have set foot in.

The Genomic Era

14 Regardless of the direction in which the Human Genome Project will take humans in the future, it has already ushered in the Genomic Era, and there is no turning back. One comfort lies in the fact that the completion of the Human Genome Project is really only the beginning of a long and uncertain journey of studying, **interpreting**, and applying the information it has amassed. How wisely that information is applied, or not applied, will determine the Human Genome Project's real value.