# UNIT 3 BASICS OF HUMAN GENOME PROJECT

**Contents**



## 3.0 OBJECTIVES

- To have an over-all understanding of Human Genome Project
- To see some of its advantages and dangers.
- To have a general idea of the technological growth that has made Human Genome Project possible.

## 3.1 INTRODUCTION

The Human Genome Project ranks right up there at the top of the scale of scientific advances. The opportunity to read our own instruction manual is holistic and astounding. What else in science could compete with that? This is the most important organized scientific endeavor that human kind has ever mounted. Human Genome Project has already identified many genetic abnormalities and will no doubt identify many more. New treatments and better safer treatments are likely to follow as a result. For this reason, we believe that the Human Genome Project is important scientific project of the century and probably, the most important of all time. Although it is based on the findings of many researches over many years, the efforts of the Human Genome Project have the potential for creating better humanity than any single scientific venture in world history.

For every good that a technology can bring to society, there is also a potential for abuse. Ethical guidelines are essential to the success of the Human Genome Project. This scientific paper gives the overall picture of Human Genome project with all its pros and cons. It not only deals with the scientific methodologies involved in the human Genome Project but also on the philosophical implication which is the mail concern. We will see in the paper how the Human Genome

Project is useful in many ways and also can be misused in many ways. Although the Human Genome Project is a great achievement in Science and especially in Genetics, there is a need for Ethical, social and moral concern. Just as dynamite which was discovered in a good intention to break the rocks were eventually misused to kill people so also the Human Genome Project which was initially a knowledge seeking project, which aimed to study the genetic nature of human being later could be misused in many ways due to the unethical , immoral and anti-social elements. Any venture must uphold the human dignity and must not dehumanize. Any sort of oppressive structures in the scientific world should be counterattacked or guided by proper ethics so that every development is based on the welfare of the whole of human race. Thus we come to a critical analysis and reflection so that we can decide for ourselves and let others know what good things we can take from this and what misuses we can avoid for the welfare of the whole of humanity at large.

Human Genome Project, international scientific collaboration that seeks to understand the entire genetic blueprint of a human being.This genetic information is found in each cell of the body, encoded in the chemical deoxyribonucleic acid (DNA). Through a process known as sequencing, the Human Genome Project has identified nearly all of the estimated 31,000 genes in the nucleus of a human cell. The project has also mapped the location of these genes on the 23 pairs of human chromosomes, the structures containing the genes in the cell's nucleus.

The data derived from mapping and sequencing the human genome will help scientists associate specific human traits and inherited diseases with particular genes at precise locations on the chromosomes. This advance will help provide an unparalleled understanding of the fundamental organization of human genes and chromosomes. Many scientists believe that the Human Genome Project has the potential to revolutionize both therapeutic and preventive medicine by providing insights into the basic biochemical processes that underlie many human diseases.

The idea of undertaking a coordinated study of the human genome arose from a series of scientific conferences held between 1985 and 1987. The Human Genome Project began in earnest in the United States in 1990 with the expansion of funding from the National Institute of Health (NIH) and the Department of Energy (DOE). One of the first directors of the U.S. program was American biochemist James Watson, who in 1962 shared the Nobel Prize for physiology or medicine with British biophysicists Francis Crick and Maurice Wilkins for the discovery of the structure of DNA. Many nations have official human genome research programs as part of this collaboration, including the United Kingdom, France, Germany, and Japan. In a separate project intended to speed up the sequencing process and commercialize the results, Celera Genomics, a privately funded Biotechnology company, used a different method to assemble the sequence of the human genome. Both the public consortium and Celera Genomics completed the first phase of the project, and they each published a draft of the human genome simultaneously, although in separate journals, in February 2001. Scientists from the public consortium completed the final sequencing of the human genome in April 2003.

## 3.2 HISTORY OF HGP

The idea of the HGP was initiated in 1977, when simple and efficient methods for sequencing DNA were described. Before that time the possibility of sequencing

the entire human genome was no more than extreme wishful thinking. In the 1980's it was becoming increasingly apparent to many scientists that an understanding of basic biology would be greatly enhanced if the detailed structure of DNA was understood. Over the last two decades, automated DNA sequencers have made the process of obtaining the base-by-base sequence of DNA easier. In 1984, for the first time a meeting was sponsored by the Department of Energy (DOE) to address the problem of detecting extremely low levels of very rare changes in DNA (mutations) in humans exposed to radiation and other environmental hazards (Postiglione and Brungs 1993). At that time, it was realized that the level of effort including the automation of DNA analysis techniques would be similar to the requirements for sequencing the human genome. Several other meetings followed until the first formal proposal appeared in 1986 published by Renato Dulbecco who focused on potential benefits to cancer research from the availability of the complete genomic sequence (McConkey 1993). The immediate public response was considerable skepticism about the possibility and economical feasibility of the HGP, the value of the results, its impact on the rest of biological research, goal definitions, funding, and potential risks of information abuse.

As a $3 billion project, it was a 15-year effort to find the estimated 80,000-100,000 human genes and determine the sequence of the 3-billion chemical bases that make up human DNA and underlies all life's diversity.

In 1976, the genome of the RNA virus Bacteriophage MS2 was the first complete genome to be determined, by Walter Fiers and his team at the University of Ghent (Ghent, Belgium). The idea for the shotgun technique came from the use of an algorithm that combined sequence information from many small fragments of DNA to reconstruct a genome. This technique was pioneered by Frederick Sanger to sequence the genome of the Phage Ö-X174, a virus that primarily infects bacteria that was the first fully sequenced genome (DNA-sequence) in 1977. The technique was called shotgun sequencing because the genome was broken into millions of pieces as if it had been blasted with a shotgun. In order to scale up the method, both the sequencing and genome assembly had to be automated, as they were in the 1980s.

Those techniques were shown applicable to sequencing of the first free-living bacterial genome (1.8 million base pairs) of *Haemophilus influenza* in 1995 and the first animal genome. It involved the use of automated sequencers, longer individual sequences using approximately 500 base pairs at that time. Paired sequences separated by a fixed distance of around 2000 base pairs which were critical elements enabling the development of the first genome assembly programs for reconstruction of large regions of genomes. Three years later, in 1998, the announcement by the newly-formed Celera Genomics that it would scale up the pair wise end sequencing method to the human genome was greeted with skepticism in some circles. The shotgun technique breaks the DNA into fragments of various sizes, ranging from 2,000 to 300,000 base pairs in length, forming what is called a DNA "library". Using an automated DNA sequence, the DNA is read in 800bp lengths from both ends of each fragment. Using a complex genome assembly algorithm and a supercomputer, the pieces are combined and the genome can be reconstructed from the millions of short, 800 base pair fragments. The success of both the public and privately funded effort hinged upon a new, more highly automated capillary DNA sequencing machine, called the Applied Bio

systems 3700, that ran the DNA sequences through an extremely fine capillary tube rather than a flat gel. Even more critical was the development of a new, larger-scale genome assembly program, which could handle the 30–50 million sequences that would be required to sequence the entire human genome with this method. At the time, such a program did not exist (Davies 2001).

One of the first major projects at Celera Genomics was the development of this assembler, which was written in parallel with the construction of a large, highly automated genome sequencing factory. Development of the assembler was led by Brian Ramos. The first version of this assembler was demonstrated in 2000, when the Celera team joined forces with Professor Gerald Rubin to sequence the fruit fly Drosophila melanogaster using the whole-genome shotgun method. At 130 million base pairs, it was at least 10 times larger than any genome previously shotgun assembled. One year later, the Celera team published their assembly of the three billion base pair human genome. This project is closely associated to the branch of biology called Bio-informatics. The human genome project international consortium announced the publication of a draft sequence and analysis of the human genome the genetic blueprint for the human being. An American company Celera, led by Craig Venter and the other huge international collaboration of distinguished scientists led by Francis Collins, director, National Human Genome Research Institute, U.S., both published their findings (Zweiger 2003).

This Mega Project is co-ordinated by the U.S. Department of Energy and the National Institute of Health. During the early years of the project, the Welcome Trust (U.K.) became a major partner, other countries like Japan, Germany, China and France contributed significantly. The two factors that made this project a success is:

1) Genetic Engineering Techniques, with which it is possible to isolate and clone any segment of DNA.

2) Availability of simple and fast technologies, to determining the DNA sequences.

Being the most complex organisms, human beings was expected to have more than 100,000 genes or combination of DNA that provides commands for every characteristics of the body. Instead their studies show that humans have only 30,000 genes – around the same as mice, three times as many as flies, and only five times more than bacteria. Scientist told that not only are the numbers similar, the genes themselves, baring a few, are alike in mice and men. In a companion volume to the Book of Life, scientists have created a catalogue of 1.4 million single-letter differences, or single-nucleotide polymorphisms (SNPs) – and specified their exact locations in the human genome. This SNP map, the world's largest publicly available catalogue of SNP's, promises to revolutionize both mapping diseases and tracing human history.

The sequence information from the consortium has been immediately and freely released to the world, with no restrictions on its use or redistribution. The information is scanned daily by scientists in academia and industry, as well as commercial database companies, providing key information services to bio-technologists. Already, many genes have been identified from the genome

sequence, including more than 30 that play a direct role in human diseases. By dating the three millions repeat elements and examining the pattern of interspersed repeats on the Y-chromosome, scientists estimated the relative mutation rates in the X and the Y chromosomes and in the male and the female germ lines. They found that the ratio of mutations in male vs female is 2:1. Scientists point to several possible reasons for the higher mutation rate in the male germ line, including the fact that there are a greater number of cell divisions involved in the formation of sperm than in the formation of eggs (Sloan 2000).

**Check Your Progress I**

**Note:** Use the space provided for your answers.s.

1) When did HGP start?

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2) Give two factors that made this project a success?

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## 3.3 HUMAN GENOME PROJECT: AN OVERVIEW

A genome is the complete collection of an organism's genetic material. The human genome is composed of about 31,000 genes located on the 23 pairs of chromosomes in a human cell. A single human chromosome may contain more than 250 million DNA base pairs, and scientists estimate that the entire human genome consists of about 3 billion base pairs.

The first steps in the Human Genome Project are to develop the needed technologies, then to „map" and „sequence" the genome. But in a sense, these well-publicized efforts aim only to provide the raw material for the next, longer strides. The ultimate goal is to exploit those resources for a truly profound molecular-level understanding of how we develop from embryo to adult, what makes us work, and what causes things to go wrong. The benefits to be reaped stretch the imagination. In the offing is a new era of molecular medicine characterized not by treating symptoms, but rather by looking to the deepest causes of disease. Rapid and more accurate diagnostic tests will make possible earlier treatment for countless maladies. Even more promising, insights into genetic susceptibilities to disease and to environmental insults, coupled with preventive therapies, will thwart some diseases altogether (Podimattom 2002).

New, highly targeted pharmaceuticals, not just for heritable diseases, but for communicable ailments as well, will attack diseases at their molecular foundations. And even gene therapy will become possible, in some cases actually "fixing" genetic errors. All of this is in addition to a new intellectual perspective on whom we are and where we came from. Begun formally in 1990, the U.S. Human Genome Project was a 13-year effort coordinated by the U.S. Department of Energy and the National Institute of Health. The project originally was planned to last 15 years, but rapid technological advances accelerated the completion date to 2003. More than 1100 top level scientists from over 18 outstanding research centers spread over 6 nations, participated in this mega project.

Francis Collins was the director of this venture. Later in 1999 Craig Venter, an eminent scientist joined and did a super-fast radical approach of short gun cloning rather than the orderly linear sequencing. The working draft DNA sequence and the more polished 2003 version represent an enormous achievement, akin in scientific importance, some say, to developing the periodic table of elements. And, as in most major scientific advances, much work remains to realize the full potential of the accomplishment. The genome is an organism's complete set of DNA. Genomes vary widely in size: the smallest known genome for a free-living organism (a bacterium) contains about 600,000 DNA base pairs, while human and mouse genomes have some 3 billion. Except for mature red blood cells, all human cells contain a complete genome.

DNA in the human genome is arranged into chromosomes—physically separate molecules that range in length from about 50 million to 250 million base pairs. A few types of major chromosomal abnormalities, including missing or extra copies or gross breaks and rejoining (translocations), can be detected by microscopic examination. Most changes in DNA, however, are more subtle and require a closer analysis of the DNA molecule to find perhaps single-base differences (Brungs 1993).

Each chromosome contains many genes, the basic physical and functional units of heredity. Genes are specific sequences of bases that encode instructions on how to make proteins. Genes comprise only about 2% of the human genome; the remainder consists of non-coding regions, whose functions may include providing chromosomal structural integrity and regulating where, when, and in what quantity proteins are made.

Although genes get a lot of attention, it's the proteins that perform most life functions and even make up the majority of cellular structures. Proteins are large, complex molecules made up of smaller subunits called amino acids. Chemical properties that distinguish the 20 different amino acids cause the protein chains to fold up into specific three-dimensional structures that define their particular functions in the cell.

The constellation of all proteins in a cell is called its proteome. Unlike the relatively unchanging genome, the dynamic proteome changes from minute to minute in response to tens of thousands of intra- and extracellular environmental signals. A protein's chemistry and behavior are specified by the gene sequence and by the number and identities of other proteins made in the same cell at the same time and with which it associates and reacts. Studies to explore protein

structure and activities, known as proteomics, will be the focus of much research for decades to come and will help elucidate the molecular basis of health and disease (Singh 2002).

## 3.4 GOALS OF HGP

The sequence of the human DNA is stored in databases available to anyone on the Internet. The U.S. National Centre for Biotechnology Information house the gene sequence in a database known as Gen Bank, along with sequences of known and hypothetical genes and proteins. Other organizations such as the University of California, Santa Cruz, and Ensemble present additional data and annotation and powerful tools for visualizing and searching it. Computer programs have been developed to analyze the data, because the data itself is difficult to interpret without such programs.

The process of identifying the boundaries between genes and other features in raw DNA sequence is called genome annotation and is the domain of bioinformatics. While expert biologists make the best annotators, their work proceeds slowly, and computer programs are increasingly used to meet the high-through put demands of genome sequencing projects. The best current technologies for annotation make use of statistical models that take advantage of parallels between DNA sequences and human language, using concepts from computer science such as formal grammars. Another, often overlooked, goal of the HGP is the study of its ethical, legal, and social implications. It is important to research these issues and find the most appropriate solutions before they become large dilemmas whose effect will manifest in the form of major political concerns.

All humans have unique gene sequences. Therefore the data published by the HGP does not represent the exact sequence of each and every individual's genome. It is the combined reference genome of a small number of anonymous donors. The HGP genome is a scaffold for future work in identifying differences among individuals. Most of the current effort in identifying differences among individuals involves single-nucleotide polymorphisms and the Hap-Map. The Human Genome Project (HGP) was an international scientific research project with a primary goal to determine the sequence of chemical base pairs which make up DNA.

The project began in 1990 and was initially headed by James D. Watson at the U.S. National Institutes of Health. A working draft of the genome was released in 2000 and a complete one in 2003, with further analysis still being published. A parallel project was conducted outside of government by the Celera Corporation. Most of the government-sponsored sequencing was performed in universities and research centers from the United States, the United Kingdom, Japan, France, Germany, China, India, Canada, and New Zealand. The mapping of human genes is an important step in the development of medicines and other aspects of health care.

While the objective of the Human Genome Project is to understand the genetic makeup of the human species, the project has also focused on several other non-human organisms such as E.coli, the fruit fly, and the laboratory mouse. It remains one of the largest single investigational projects in modern science. The Human Genome Project originally aimed to map the nucleotides contained in a human

haploid reference genome. Several groups have announced efforts to extend this to diploid human genomes including the International Hap Map Project. The "genome" of any given individual (except for identical twins and cloned organisms) is unique; mapping "the human genome" involves sequencing multiple variations of each gene. The project did not study the entire DNA found in human cells; some heterochromatic areas remain un-sequenced.

## 3.5 ADVANTAGES OF HUMAN GENOME PROJECT

The main advantages of HGP could be seen two: (i) Knowledge of the effects of variation of DNA among individuals can revolutionize the ways to diagnose, treat and even prevent a number of diseases that affects the human beings. (ii) Providing clues to the understanding of human biology.

***Diagnosing and Predicting Disease and Disease Susceptibility***

All diseases have a genetic component, whether inherited or resulting from the body's response to environmental stresses like viruses or toxins. The successes of the HGP have even enabled researchers to pinpoint errors in genes—the smallest units of heredity—that cause or contribute to disease. The ultimate goal is to use this information to develop new ways to treat, cure, or even prevent the thousands of diseases that afflict humankind. But the road from gene identification to effective treatments is long and fraught with challenges. In the meantime, biotechnology companies are racing ahead with commercialization by designing diagnostic tests to detect errant genes in people suspected of having particular diseases or of being at risk for developing them.

An increasing number of gene tests are becoming available commercially, although the scientific community continues to debate the best way to deliver them to the public and medical communities that are often unaware of their scientific and social implications. While some of these tests have greatly improved and even saved lives, scientists remain unsure of how to interpret many of them. Also, patients taking the tests face significant risks of jeopardizing their employment or insurance status. And because genetic information is shared, these risks can extend beyond them to their family members as well.

Hazards of Human Genome Project: We also need to be aware of the various dangers we are exposing ourselves into, while pursuing such a venture. This will be taken up in the next units.

***Bioinformatics***

The completed human genome sequence generated a catalog made up of around 31,000 human genes; high-resolution maps of the chromosomes, including hundreds of thousands of landmarks; and billions of base pairs of DNA-sequence information. Laboratory information-management systems, robotics, database-management systems, and graphical user interfaces were among the computing tools required to help genome researchers make sense of this flood of data. A new field of research, bioinformatics, has developed in part to address the computing challenges raised by the project. Researchers in bioinformatics have developed public databases connected to the Internet to make genome data available to scientists worldwide, along with analytical software for making sense

of this flood of biological information. For example, DNA-sequence information is stored in several databases, including the NIH's Gen Bank, the European Molecular Biology Laboratory's Nucleotide Sequence Database, and the DNA Data bank of Japan.

**Check Your Progress I**

**Note:** Use the space provided for your answers.s.

1) Mention some of the advantages of HGP?

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2) What is Bioinformatics?

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## 3.6 ACHIEVEMENT OF HUMAN GENOME PROJECT

In April 2003, researchers announced that the Human Genome Project had completed a high-quality sequence of essentially the entire human genome. This sequence closed the gaps from a working draft of the genome, which was published in 2001. It also identified the locations of many human genes and provided information about their structure and organization. In addition to the human genome, the Human Genome Project sequenced the genomes of several other organisms, including brewers' yeast, the roundworm, and the fruit fly. In 2002, researchers announced that they had also completed a working draft of the mouse genome. By studying the similarities and differences between human genes and those of other organisms, researchers can discover the functions of particular genes and identify which genes are critical for life.

## 3.7 HGP: FUTURE PROSPECTS

The benefits of Human Genome Project research include the improvements in medicine, microbial genome research for fuel and environmental cleanup, DNA forensics, improved agriculture and livestock, better understanding of evolution and human migration, and more precise and accurate risk assessment.

Our knowledge about human genetics clearly expands at a great rate over the coming years. This fundamental understanding will permit control over many

biological processes, and biological control will transform medicine, agriculture, animal husbandry, and pharmaceutical production. The project has already stimulated significant investment by large corporations and lead to the creation of new companies hoping to capitalize on the project's profound and inestimable implications. Great desire exists among Biotechnology companies to acquire efficient technologies such as the genome-driven drug discovery. An understanding of human DNA certainly will be an important key in understanding a host of human diseases. Cancers, in particular, are now being understood as genetic diseases, since cancerous growths arise from either acquired or hereditary changes in cellular DNA (Postiglione and Brungs 1993). Once we know how altered DNA induces cancer development, effective tools can be developed to prevent or treat malignant growths. It is important that this knowledge will be used well, and not to stigmatize or discriminate, but to improve human health.

The HGP should illuminate fundamental functions of the body and become invaluable basis for genomic technology; however it will primarily open a fascinating area for exploration. A large portion of the value of the projects rests on the expansion of our basic understanding of biological life in general and the explicit promise of the relief of suffering from the more than 4,000 genetic hereditary diseases (i.e. Huntington disease and cystic fibrosis) either through prevention or cure.

- **Medicine:** improved diagnosis of disease. The HGP will accelerate the acquisition of probes for genes that determine an individual's susceptibility to heart disease, to certain types of cancer, to diabetes, and to some types of mental illness. We expect to learn the underlying causes of many genetic diseases, including sickle cell anemia, Huntington disease, cystic fibrosis, and several forms of cancer. This will enable us to predict the likelihood of the disease occurrence in any individual.

- **Microbial Research:** new energy sources, bio fuels.

- **DNA Forensics:** identifying potential suspects at a crime scene.

- **Agriculture:** more nutritious produce.*

- **Evolution and Human Migration:** study migration of different population groups based on female genetic inheritance.

- **Risk Assessment:** reduce the likelihood of heritable mutations and cure. Understanding of the human genome will have an enormous impact on the ability to assess risk posed to individuals by exposure to toxic agents and scientists know that genetic differences make some people more susceptible and others more resistant to such agents. Far more research work will be needed to determine the genetic basis of variability. This knowledge will help us to understand the effects of low level exposures to radiation and other energy-related agents, especially in terms of cancer risk.

The advantage of the Human Genome Project has been the recognition that it attracted extra funding to the work, raised the profile of the effort within the scientific communities, and provided elements of organization and cooperation that would not have occurred with individual scientists pursuing projects based on their personal interest. Knowledge gained by such efforts as the human genome

project will help enable experts to analyze individual differences among genomes (Zweiger 2003). Because genetic variations cause or contribute to certain diseases, we need to be aware of the dangers and hazards that HGP and Genetic Research can cause. Analysis of a person's genome could reveal health information that should remain private. People are concerned of preventing the misuse of such information and the hazardous side-effects of Genetic Research.

## 3.8 PHILOSOPHICAL REFLECTIONS

Throughout the age's human has struggled with the subject of right and wrong, ethics and justice. Ethics consists of the actions an individual takes on for oneself. No matter how criminal an individual is, he will be trying, one way or another, to put ethics on himself. The nature of the human person is the basic criterion in deciding upon ethics. Aristotle (384-322 B.C.) also got involved with ethics. He explained unethical behavior by saying that humans' rationality became over ruled by his desire. Ethics consists basically of rationality towards the highest level of survival for the individual, family, group, mankind and the environment collectively. Ethics is a reason and the smartest solution to any problem is that solution which creates the greatest good for the greatest number. Any solution that falls short of this model contains weaker reasoning. Survival is not merely the barest necessities of life; it is a graduated scale with pain and death at the bottom and immortality at its top. Everyone has an infinite ability to survive. How well one accomplishes this is depended on how well one applies ethics to life. Ethical actions are survival actions. Know that the fundamental principal of existence is to survive. Evil, illness, misfortune, and decay go hand in hand, all are the fruits of one's misdeeds!

The nature of the human person has to be considered seriously. The technologies and scientific advancements are for the welfare of the human society. There is inequality in this world based on money, race, sex, and caste. But the underlying principle of humankind is the human nature which is uphold by many religions. Even some religions do not allow the women, children to be treated equally as men. The frame of reference of some religions, that is the scriptures, bring out inequality in seeing the same fellow human beings under the banner of the caste and out caste.

Media again projects the human being as a sexual, luxurious and dreaming being which has no relevance to the existential reality. Science, Religion and Society seem to take a different route in their journey. In this situation there has to be a common understanding of taking the human person seriously with the core importance given to the poor and the rich, the learned and the illiterate, the black and the white etc. All need to understand that they are in one cosmos and sharing the same existence. Scientism and religious fanaticism has to be dealt with some concrete ways as it misleads people to become more oppressive and dehumanizing.

Goodness and advancements have to be taken as an overall welfare or affair which is a necessary one. All these years without much genetic knowledge people have been living with harmony. Hitler came to improve his race. With the advent of Human genome project, the epiphany of its misuse is already known to us. Pain and suffering has been ruling the world and humanity in so many ways. Humanity in the form of advancement is thinking newly, differently and

independently for the welfare of its future. What is welfare for some becomes horror news for the others. Well-wishers of humanity have to think in a more liberative way so as to bring in a constructive reality which will unify all peoples – where the dignity of human kind preserved and maintained with its utmost care.

Human beings are end in themselves and they are not mere means. Trans Human Species: Some scientists have proposed that there could be a trans-human species (Homo sapiens super) would emerge due to genetic manipulation which will eventually look down upon the Homo sapiens. What would be the future of the present Homo sapiens?

Beyond the moral significance of the project itself, the human genome project does indeed raise many interesting and challenging questions, questions related to the use of the tests and information it will produce. For example, it will be necessary to consider the ways in which resultant genetic probes should be used in matter of employment, insurability, money lending, reproduction, counseling and so on.

One of the major expectations of the genome project is that its information will offer people better health. But genetic characterizations are one thing and successful medical interventions to correct genetic defeccts are quite another. The goal of the genome project is to produce a characterization of the human genetic complementation in the way that anatomy produces a representation of the structural components of the human body or in the way that physiology represents bodily function. Thus genomic characterization will not identify the genome as a single person any more than anatomical or skeletal characterizations representation a given individual.

French philosopher and mathematician Blaise Pascal wrote in an essay “Prayer to ask God for the use of sickness”. The title here is problematic to contemporary consciousness. What could thus signify? That if there is a right use of sickness, there is also a wrong use of sickness? What is the purpose to sickness at all? However strange that the question might appear today, in Pascal’s view, sickness could be put to the use of personal transformation and was useful in guiding one to correct moral priorities.

What is the right use of the human genome project? Will it be used to prop the existing scientific status quo and perhaps thereby impede the aims of science? Will it be used in a campaign against difference, or will it be used to map the fullness and plenitude of existence? Will it be used as a stratagem to create a new kind of inferiority? Or will we able to understand the way in which genomic characterizations represent one possible map of a small corner of the vastness of existence? Will the goal of biomedicine be the leveling of all genetic difference in order to accommodate the social requirements of the time? Even as it offers some answers, science also creates uncertainty; even as it conquers some social evils, it also may cause evasion of social problems.

**Check Your Progress III**

**Note:** Use the space provided for your answers.s.

1) What is risk assessment?

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2) In the light of the research on HGP, do you consider that sickness has a positive value?

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## 3.9 LET US SUM UP

In this unit we have studied the basic issues connected with HGP in a very general and descriptive manner. We have not dealt with its disadvantages and hazards, but focussed on its achievements and advantages.

## 3.10 KEY WORDS

**Bioinformatics** : Bioinformatics is the application of statistics and computer science to the field of molecular biology. It is used extensively in Human Genome Project.

**E. coli or Escherichia coli** : A bacillus *or a type of bacteria* normally found in the human gastrointestinal tract and existing as numerous strains, some of which are responsible for diarrheal diseases. Other strains have been used experimentally in molecular biology.

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**On-line Resources**

Forensics: forensics, *www.ornl.gov/hgmis/elsi*/forensics.*shtml*

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The U.S. Department of Energy Office of Science, Office of Biological and Environmental Research, *Human Genome Program*, genomics.energy.gov.6-10-2010.