
UNIT 1 INTRODUCTION AND OVERVIEW OF THE COURSE

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1.0 OBJECTIVES

- To have an introduction and over-all perspective of philosophy of technology.
 - To see the larger implications of technology and philosophy of technology.
 - To see the relationship between science and technology
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1.1 INTRODUCTION

If philosophy is the attempt “to understand how things in the broadest possible sense of the term hang together in the broadest possible sense of the term”, (Sellars 1962), philosophy should not and cannot ignore technology. It is largely by technology that contemporary society hangs together. It is hugely important not only as an economic force but also as a cultural force. During the last two centuries, much philosophy of technology has been concerned with the impact of technology on society (Franssen, Lokhorst and van de Poel 2011). This unit starts with a brief historical overview, then presents an introduction to the modern philosophy of technology, and ends with a discussion of the societal and ethical aspects of technology. Then we see the course outline and its rationale.

1.2 HISTORICAL DEVELOPMENTS

It may be claimed that philosophical reflection on technology is about as old as philosophy itself. It started in ancient Greece. One early theme is the thesis that technology learns from or imitates nature (Plato). According to Democritus, for example, house-building and weaving were first invented by imitating swallows and spiders building their nests and nets, respectively. Aristotle referred to this tradition by repeating Democritus' examples, but he did not maintain that technology can only imitate nature: “generally art in some cases completes what nature cannot bring to a finish, and in others imitates nature” (Franssen, Lokhorst and van de Poel 2011)

Another theme related to philosophy of nature is the claim that there is a fundamental ontological distinction between natural things and artifacts. According to Aristotle the former have their principles of generation and motion inside, whereas the latter, insofar as they are artifacts, are generated only by outward causes, namely human aims and forms in the human soul. Natural products (animals and their parts, plants, and the four elements) move, grow, change, and reproduce themselves by inner final causes; they are driven by purposes of nature. Artifacts, on the other hand, cannot reproduce themselves. Without human care and intervention, they vanish after some time by losing their artificial forms and decomposing into (natural) materials. For instance, if a wooden bed is buried, it decomposes to earth or changes back into its botanical nature by putting forth a shoot. The thesis that there is a fundamental difference between man-made products and natural substances had a long-lasting influence. In the Middle Ages, Avicenna criticized alchemy on the ground that it can never produce ‘genuine’ substances. Even today, some still maintain that there is a difference between, for example, natural and synthetic vitamin C. Aristotle's doctrine of the four causes—material, formal, efficient and final—can be regarded as a third early contribution to the philosophy of technology. Aristotle explained this doctrine by referring to technical artifacts such as houses and statues (Wikipedia). A final point that deserves mentioning is the extensive employment of technological images by Plato and Aristotle. In his “Timaeus,” Plato described the world as the work of an Artisan, the Demiurge. His account of the details of creation is full of images drawn from carpentry, weaving, modelling, metallurgy, and agricultural technology. Aristotle used comparisons drawn from the arts and crafts to illustrate how final causes are at work in natural processes. Despite their criticism of the life led by merely human artisans, both Plato and Aristotle found technological imagery indispensable for expressing their belief in the rational design of the universe. Considered under the rubric of the Greek term *techne* (art, or craft knowledge), the philosophy of technology goes to the very roots of Western philosophy. In his “Republic,” Plato sees *techne* as the basis for the philosophers' proper rule in the city. In the “Nicomachean Ethics,” Aristotle describes *techne* as one of the four ways that we can know about the world. The Stoics argued that virtue is a kind of *techne* based upon a proper understanding of the universe.

Some prominent 20th century philosophers to directly address the effects of modern technology on humanity were John Dewey, Martin Heidegger, Herbert Marcuse, Günther Anders and Hannah Arendt. They all saw technology as central to modern life, although Heidegger, Anders, Arendt and Marcuse were more ambivalent and critical than Dewey. The problem for Heidegger was the hidden nature of technology's essence, *Gestell* or Enframing which poised for humans what he called its greatest danger thus its greatest possibility. Heidegger's major work on technology is found in “The Question Concerning Technology.” He argues that technology is not just a tool used by humans but it has become an overarching frame, which essentially makes up humans.

1.3 DIFFERENT FIELDS OF PHILOSOPHY OF TECHNOLOGY

The vast field of philosophy of technology can treat any or all of the following areas of study.

- Critique of technology
- Ethics of technology
- History of technology
- Industrial sociology

- Philosophy of engineering
- Technological evolution
- Theories of technology
- Analytic Philosophy of Technology

These different fields of study give us an idea of the various dimensions of philosophy of technology and its relevance.

Check Your Progress I

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

1) What is “techne”?

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2) Give the different fields of philosophy of technology?

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1.4 THE RELATIONSHIP BETWEEN TECHNOLOGY AND SCIENCE

The close relationship between the practices of science and technology may easily keep the important differences between the two from view. The predominant position of science in the philosophical perspective did not easily lead to a recognition that technology merited special attention for involving issues that did not emerge in science. This situation is often presented, perhaps somewhat dramatized, as coming down to a claim that technology is ‘merely’ applied science (Franssen, Lokhorst and van de Poel 2011).

A questioning of the relation between science and technology was the central issue in one of the earliest discussions among analytic philosophers of technology. In 1966, in a special issue of the journal “Technology and Culture,” the Polish Philosopher, Henryk Skolimowski, argued that technology is something quite different from science. As he phrased it, science concerns itself with what is, whereas technology concerns itself with what is to be. A few years later, in his well-known book “The sciences of the artificial” (1969), Herbert Simon, an American political scientist, emphasized this important distinction in almost the same words, stating that the scientist is concerned with how things are but the engineer with how things ought to be. Although it is difficult to imagine that earlier philosophers of science were blind to this difference in orientation, their inclination to view knowledge as a system of statements may have led to a conviction that in technology no knowledge claims play a role that cannot also be found in science.

In the same issue of “Technology and Culture,” Mario Bunge, an Argentine philosopher and physicist, defended the view that technology is applied science, but in a subtle way that does justice to the differences between science and technology. Bunge acknowledges that technology is about action, but an action heavily underpinned by theory—that is what distinguishes technology from the arts and crafts and puts it on a par with science. According to Bunge, theories in technology come in two types: substantive theories, which provide knowledge about the object of action, and operative theories, which are concerned with action itself. The

substantive theories of technology are indeed largely applications of scientific theories. The operative theories, in contrast, are not preceded by scientific theories but are born in applied research itself. Thus, as Bunge holds, operative theories show a dependency on science in that in such theories the method of science is employed. (Franssen, Lokhorst and van de Poel 2011).

1.5 ETHICAL AND SOCIAL ASPECTS OF TECHNOLOGY

Neutrality Thesis

It was not until the twentieth century that the development of the ethics of technology as a systematic and more or less independent subdiscipline of philosophy began to take shape. This late development may seem surprising given the large impact that technology has had on society, especially since the industrial revolution (Franssen, Lokhorst and van de Poel 2011).

A plausible reason for this late development of ethics of technology is the instrumental perspective on technology. This perspective implies, basically, a positive ethical assessment of technology: technology increases the possibilities and capabilities of humans, which seems in general desirable. Of course, since antiquity, it has been recognized that the new capabilities may be put to bad use or lead to human hubris. Often, however, these undesirable consequences are attributed to the users of technology, rather than the technology itself, or its developers. This vision is known as the instrumental vision of technology resulting in the so-called neutrality thesis. The neutrality thesis holds that technology is a neutral instrument that can be put to good or bad use by its users. During the twentieth century, this neutrality thesis met with severe critique, most prominently by Martin Heidegger and Jacques Ellul, who have been mentioned in this context, but also by philosophers from the Frankfurt School (Adorno, Horkheimer, Marcuse, Habermas). Today most philosophers do not subscribe to the neutrality thesis, since technology is not always in itself neutral.

Ethics of specific technologies

The last decades have witnessed an increase in ethical inquiries into specific technologies. One of the most visible new fields is probably computer ethics, but biotechnology has spurred dedicated ethical investigations as well. Also more traditional fields like architecture and urban planning have attracted specific ethical attention. More recently, nanotechnology and so-called converging technologies have led to the establishment of what is called nanoethics. Apart from this, there has been a debate over the ethics of nuclear deterrence (Franssen, Lokhorst and van de Poel 2011).

Obviously the establishment of such new fields of ethical reflection is a response to social and technological developments. Still, the question can be asked whether the social demand is best met by establishing new fields of applied ethics. This issue is in fact regularly discussed as new fields emerge. Several authors have, for example argued that there is no need for nanoethics because nanotechnology does not raise any really new ethical issues. The alleged absence of newness here is supported by the claim that the ethical issues raised by nanotechnology are a variation on, and sometimes an intensification of, existing ethical issues, but hardly really new, and by the claim that these issues can be dealt with the existing theories and concepts from moral philosophy. Thus every new technology creates its own specific ethical issues which are usually addressed in philosophy of technology.

Responsibility

Responsibility has always been a central theme in the ethics of technology. The traditional philosophy and ethics of technology, however, tended to discuss responsibility in rather general

terms and were rather pessimistic about the possibility of engineers to assume responsibility for the technologies they developed. The French philosopher and law professor, Jacques Ellul, for example, has characterized engineers as the high priests of technology, who cherish technology but cannot steer it. In engineering ethics, the responsibility of engineers is often discussed in relation to code of ethics that articulate specific responsibilities of engineers. Such codes of ethics stress three types of responsibilities of engineers: 1) conducting the profession with integrity and honesty and in a competent way, 2) responsibilities towards employers and clients and 3) responsibility towards the public and society. With respect to the latter, most US codes of ethics maintain that engineers 'should hold paramount the safety, health and welfare of the public'. The larger issue of the responsibility technology has for the human welfare needs to be addressed both generally and in its particular fields. So ethical, social and even legal issues arising out of emerging technologies is definitely part of philosophy of technology.

1.6 PHILOSOPHIZING AS A SEARCH

In this section we briefly try to relate philosophizing and technology. Wolfgang Stegmüller, a German philosopher, distinguishes three different kinds of practising philosophy.

1. Philosophy as *Weltanschauung* (world-view). The aim is either to enhance (or replace) religion or at least to give some advice to master the difficulties of life. (Marxism, Existentialism)
2. Philosophy as science that aims to acquire theoretical knowledge, either independent on the natural sciences or expending them and approaching a kind of overall view or "*Gesamtschau*" (Brentano, Husserl, Hartmann, Schelers)
3. Philosophy as investigation of the fundamentals of the empirical and theoretical sciences (Vienna Circle and analytical philosophy)

Whatever be the view of philosophy we hold, we cannot avoid technology and its overall effects in our philosophizing. In this course, our aim is not to study some of the significant technological innovations that contemporary society is facing and to dialogue with them critically and creatively. We believe that some of the emerging technological fields contribute much to the growth of human understanding and philosophical reflection.

In general we may speak of three areas of philosophizing: the self (humanity in general), the world and God (which implies only the Transcending nature of humans is one does not believe in God).

In the overview of this course, we have been trying to see how technology can through light on each of these individual areas of philosophizing. The first unit that we have covered throws further light on the nature of reality. The second one throws light on life in general and human life in particular. The third unit that deals with consciousness and artificial intelligence is an attempt to speak more on individual identity and also on God. Neuroscience and neurotheology, while trying to understand the mysterious nature of the brain, attempts to throw light on our understanding of self and of God. The final unit takes seriously the phenomenon of death which is intrinsic to human beings. Recent claims by technology that it can overcome death radically alters the way humans understand ourselves and God.

Check Your Progress II

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

1. How does Jacques Ellul characterize engineers?

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2. Who view philosophy as a *Weltanschauung*?

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1.7 COURSE OVERVIEW AND THE RATIONALE

The way we understand the world and ourselves is to a large extent shaped by the world we live in. Or better, our world-view truly shapes our understanding of ourselves and the world. For instance, the static world view of the ancients was radically altered by Newton's model of the world as a machine. Such an understanding brought about by the scientific discoveries of Newton changed our understanding of humans, world and God also. That is the theme of the first block.

In the last century two of the greatest scientific theories have been the theory of relativity ("the most creative theory") and the theory of quantum mechanics ("the most successful theory"). Together they have altered radically the way we understand the world: our cosmology. Big Bang Theory, which is intimately connected to these two theories explain the origin of the universe. The very nature of time, space at the macro-world and micro-world that these two theories provide are quite different from the classical view of nature. Even after 100 years of their discoveries we have not really fathomed the depth of these insights regarding the nature of the empirical world.

It is in this background that we have introduced two further theories that furthers our understanding of nature. The theory of chaos and nanotechnology, which we have discussed in the previous block, proposes to us a world quite different from the classical, Newtonian or even Einsteinian worlds. These two theories through different insights into the nature of reality.

For instance the theory of chaos tells us that there is a very close connection between apparent order and chaos. The butterfly effect may point to the fact that some events in the universe may go beyond the principles of causality. The fractal nature of reality tells us that roughness in the world (not the idealisations of the world) is what really counts.

Coming to the nanotechnology, which is the science of building machines at a subatomic level, we have a lot to learn. At that realm, which is far greater than the quantum level, some substances develop different qualities. There will be tremendous revolution in our life-style brought about by nanotechnology in the immediate future. Many of these path-breaking discoveries will have philosophical implications. It changes the way we understand the world. It shatters our old world-view and ushers in a more dynamic and even extremely complex and involved nature of reality, of which we are part of.

The next block, moves from reality to life. The tremendous changes brought about by quantum mechanics (or electronics) in the last thirty years will pale away in comparison to the changes that will be brought in biotechnology in the next few decades. Quantum mechanics shattered our way of understanding the world. Biotechnology, most probably, will shatter the way we approach life, something much more intimate to us. Such changes will occur in the coming years and that is why this century is called the "century of biotechnology." The way we feel about life,

the respect and dignity we ascribe to it and the way we treat other living beings will be challenged by the latest inventions of biotechnology. Already there are talks about “artificial life” and “artificial cells.” Though they still remain at the theoretical realm, they will have quite significant repercussions on our own lives and on our understanding of life. The danger posed by such technologisation is the commodification of life. The opportunity provided by such technologies is the human ability to enhance life.

This leads us to the third block that deals still more intimate aspects of human life: our own self-identity and self-consciousness. The latest neurological findings will surely throw further light on our self-understanding, consciousness, self-identity and all those aspects that make our human life so unique. Advances in brain studies, it is hoped, will provide solace to many sick people, prevent many neurological sickness and further help us to understand ourselves. Though we are just beginning in the process of understanding our brain, it is hoped, that technological advances will improve and enhance our own self-understanding.

Closely connected with our self-understanding is our understanding of God. There are some news that some scientists have discovered the “God-spot” in the brain or that there are “spiritual neurons.” Even if we do not discover such specific God pointers, we need to appreciate the fact that it is our brain that enables us to approach and accept God. We do not believe that God is an illusion created by God. But we can rightly claim that it is the brain that enables us to experience God. The mystical experiences have a neurological basis and spirituality and the transcendental feeling of being united with the whole cosmos is enabled and stimulated by brain. Therefore, without doubt, our understanding of God will be sharpened by the advances in brain studies. That is the topic that we deal with in the third Block.

The last block deals with death. After dealing with the philosophy of death, we take up some technological marvels that claims that death may be eliminated. They are still tall claims. But many techno-enthusiasts have sold themselves to this idea. So much of money is invested into this kind of death. Though we ought to remain skeptical of such claims, we need to realize the tremendous such an idea can have on the society. The idea that death may be overcome, even when it remains merely an idea all the time, can affect our very understanding and philosophizing.

Finally we talk of the crucial possibilities open to ourselves at this point in history: cosmic extinction or collective extension. Truly humanity can destroy ourselves and the whole of life from our precious planet earth. We have the technological prowess for it. We can also positively enhance life in its totality. We do have the technology for it. At the moment we are at the cross-roads! We can either choose life or death, for ourselves and for the rest of humanity.

1.8 LET US SUM UP

In this unit, after introducing philosophy of technology, we have seen the relationship between science and technology. Then we saw the ethical dimensions of technology. Finally, we saw the rationale of our course.

Check Your Progress III

Note: Use the space provided for your answers.

1. How do you relate philosophizing to technology?

2. How does neuroscience change the way we understand God and ourselves?

1.9 KEY WORDS

Techne or **techné**: As distinguished from episteme, is etymologically derived from the Greek word τέχνη which is often translated as craftsmanship, craft, or art. It is the rational method involved in producing an object or accomplishing a goal or objective. Techne resembles *epistēmē* in the implication of knowledge of principles, although techne differs in that its intent is making or doing, as opposed to "disinterested understanding."

Gestell or **Enframing**: Gestell (or sometimes Ge-stell) is a German word used by Twentieth century German philosopher Martin Heidegger to describe what lies behind or beneath modern technology. According to him, the essence of technology is Gestell. Indeed, "Gestell, literally 'framing', is an all-encompassing view of technology, not as a means to an end, but rather a mode of human existence."

Hubris: In Greek tragedy hubris is excessive pride toward or defiance of the gods, leading to nemesis (destruction and death). This is the danger that humans are all exposed to.

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UNIT 2 GENETICS AND STEM CELL RESEARCH

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2.0 OBJECTIVES

- To have an over-all understanding of Genetics
 - To see some of its possibilities and dangers of stem-cell research.
 - To visualise how genetics will change life in the coming decades.
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2.1 INTRODUCTION

Human life today has been enormously impacted by advances in the science of genetics and related work on the physiology of human reproduction. Today human beings could be understood as “Evolution become capable of consciously extending or eliminating itself” (Pandikattu 2006). This is true with regard to genetics and its breakthrough in science. There is explosion of genetic knowledge especially in recent history has helped us to reach a position in which we can extend and eliminate ourselves. The potential applications of Genetics, Biotechnology, and Molecular biology - when used responsibly - will make valuable contributions to human society as a whole. The field of Genetics is in the news daily, with researchers mapping the human genome cloning animals, and identifying new disease genes. For many people, the problem is putting this information into context and understanding what recent genetic "breakthroughs" really mean in terms of our health and life.

The first part of this unit deals with Genetics and Genetic engineering. It would give us an understanding of what is Genetics and its applications today in our scientific field. It also deals with the history of Genetics and future prospects of Genetic engineering. In the second part we explain about the stem cell research with particular reference to the Human Genome Project. The explosion of genetic knowledge especially in recent history has helped us to reach a position in which we can extend and eliminate ourselves. Here we have a choice to make. It is a matter of conscious choice that we make- a collective, creative, coherent choice. As the book of

Deuteronomy in the Holy Bible says “I have set before you today life and prosperity, death and adversity.” Choice is ours....this choice is not for profit, not for progress, but for humanity, for life.

2.2 GENETICS AND GENETIC ENGINEERING

GENETICS AND DNA- OVER VIEW

As a scientific study of hereditary, Genetics deals with the passing on of characteristics of one generation to the next. Geneticist investigates the structure, function and transmission of genes. Genes are the basic units of hereditary information and are present in the cells of all organisms. For example, each of the cells in the human body has about 30,000 to 40,000 genes. They determine over all body build up and traits such as eye, hair and skin color. The term Genetics is a term coined by W. Bateson to designate the portion of Biology that deals with heredity, variation development and evolution. According to British geneticist J. B. S Haldane, “Genetics is the branch of Biology which is concerned with innate differences between similar organisms.....” however Genetics can be defined as the science that deals with the structure, organization, transmission and function of genes and the origin of variation in them (Singh 2002).

DEOXYRIBONUCLEIC ACID (DNA)

We human beings need to be educated to become more "DNA literate" - as our understanding of the science of Genetics is the key to its future uses. Every human cell contains a nucleus, within which lie the 46 human chromosomes that determine each individual's genetic makeup (genotype). Chromosomes are made up of DNA, which in turn is made up of nitrogenous bases. In other words, DNA (Deoxyribonucleic Acid) is a thin, long, and chainlike, molecule that is contained within almost all of our cells in a compartment called the nucleus and found in every living cell on earth. However it is important to note that DNA even occurs in bacterial cells, which do not have a nucleus and in some viruses (WBE 2004).

DNA directs the formation, growth, and reproduction of cells in the organisms. It is composed of individual units called nitrogenous bases. The DNA molecule is shaped like a twisted ladder, or double helix. The sides of this ladder are made of alternating sugar and phosphate molecules. The rungs of the ladder are made of four nitrogenous bases - guanine, cytosine, adenine, and thymine. These bases, abbreviated G, C, A, and T, respectively, and form pairs on the ladder to create a complete rung. Adenine only pairs with thymine(A-T) and guanine only pairs with cytosine(G-C). Therefore, the sequence of bases on one side of the ladder always predictably complements the sequence on the other side (Postiglione and Brungs 1993).

Check Your Progress I

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

1) What is DNA?

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2) “I have set before you today life and prosperity, death and adversity.” Comment in the light of Genetic engineering.

2.3 BRIEF HISTORY OF GENETICS

References to Genetics can be traced back to biblical times. We know from the book of Genesis, Jacob, the son of Isaac had a method by which his sheep and goats gave birth to spotted and speckled offspring (Genesis 30: 37-39).

The Babylonians had the knowledge that for a date to be fruitful, pollen from the male palm had to be introduced to the pistils of the female palm. The ancient Greek philosophers were the first to look at the world in a scientific fashion. They developed theories of everything and Genetics was no exception. Aristotle through his observation learned that the male and female do not make equal contribution to their offspring. According to him their contributions were qualitatively different: the female gives “matter” and the male gives “motion”.

The modern science of Genetics began in 1900, when the fundamental laws determining the transmission of hereditary traits from one generation to the next were discovered. We human beings carry within us “not simply not an injunction to reproduce after our kind, but to reproduce specific features of height, weight, skin color, eyes, hair and so on” (Guttman et al 2006). The fundamental laws of hereditary are laws which apply to all plants and animals as well as many microorganisms for they demonstrate similarities among life forms.

In late 1970’s, the ability to directly manipulate the genes of plants and animals was developed. Much controversy started when the proposals to begin human gene manipulation were put forth in the early 1980’s. A small number of researchers argued in favour of germ line manipulation, but the majority of scientists and others opposed it. In 1983 a letter signed by 58 religious leaders said, “Genetic engineering of the human germline represents a fundamental threat to the preservation of the human species as we know it, and should be opposed with the same courage and conviction as we now oppose the threat of nuclear extinction.” In 1985 the U.S. National Institute of Health (NIH) approved somatic gene therapy trials, but said that it would not accept proposals for germline manipulation “at present.”

Though the advocates of germline manipulation received many setbacks, germline engineering moved to the status of an openly acknowledged political cause in March 1998, when Gregory Stock, Director of the Program on Medicine, Technology and Society at the University of California at Los Angeles, organized the symposium “Engineering the Human germline.” All the speakers were avid proponents of germline engineering. Stock declared that the important question was “not if, but when” germline engineering would be used. The symposium was attended by nearly 1,000 people.

After few months of the University of California, Los Angeles(UCLA) conference one of the key participants, somatic gene transfer pioneer W. French Anderson, submitted a draft proposal to the NIH to begin somatic gene transfer experiments on human fetuses. He was of the view that this procedure could have a “relatively high” potential for “inadvertent gene transfer to the germline.” Anderson’s proposal is widely acknowledged to be strategically crafted so that approval could be construed as acceptance of germline modification, at least in some circumstances.

In 1966, the first successfully cloned sheep Dolly became the symbol of the progress of cloning and has helped the population to understand the significance of cloning for humanity. Dolly was born 5 July 1996 to three mothers (one provided the egg, another DNA and a third carried the cloned embryo to term) She was created using the technique of somatic cell nuclear transfer.

Meanwhile the official announcement of the success of Human Genome Project and the mapping of the “working draft” Human Genomes became the climax of the genetic march forward. The Human Genome Project was formally completed in April 2003. Research on development of Genetics had put us in a situation where we can transform what we are, where we are and the way we live both positively as well as negatively.

If the current pace of research and development continues, there will be an explosion of genetic knowledge and capability over the next several years. We will be able to transform the Biology of plants, animals, and people with the same detail and flexibility as today’s digital technologies and the microchip enable us to transform information. The challenge before us is to summon the wisdom, maturity, and discipline to use these powers in ways that contribute to a fulfilling, just, sustainable world, and to forgo those uses that are degrading, destabilizing and – quite literally – dehumanizing (Pandikattu 2005).

2.4 GENETICS- FUTURE PROSPECTS

Genes play an important role in shaping what we are today and for all our so-called genetic traits. The genetic information of parents is passed down to their children, grand children, or even great grand children. Similarly we have also inherited a number of genetic traits from our ancestors. All our physical characteristics, personality traits, and talents can be the result of the genetic make-up of our ancestors. We have 23 pairs of chromosomes in almost every cell in our body. Each pair consists of a chromosome from our mother and a chromosome from our father. The color of our hair, our height, and even our predisposition to health concerns are some of the genetic traits that we have inherited from our parents. Our genetic make-up is also responsible for a number of health problems that we face. Some examples of genetic, or inherited, health problems include obesity, heart disease, cancer, diabetes and hypertension. Many health concerns come about due to a combination of our inherited genetic make-up.

With the human Genome decoded, researchers have the daunting task of sifting through the newly-discovered genes in search of those that lead to disease. These efforts will change the way we view diseases and receive medical care. Once researchers know which genes are involved in a disease, they can develop a test to screen people who are at risk and also start looking for a cure. This will identify high-risk people who may require more intensive screening or preventative action. Knowing what genes cause a given disease can also help researchers

understand what goes wrong in that disease, which can help drive the search for drugs that counteract the problem.

Genomic Medicine

The genomic medicine has helped the humanity in disease gene discovery (Disease taxonomy), clinical introduction of drugs with novel therapeutic actions, more sensitive diagnostic tests, medical treatment tailored to individual genotypes, predictive testing for genetic diseases and new methods of preventive medicine. Before the flowering of genome science, disease taxonomy was captive to relatively insensitive morphological techniques (Chan and Chia 2003). The emergence of Transcription profiles has helped scientists to understand that diseases that were previously considered to be single entities possess several distinct genetic signatures.

Personalized Treatments

Personalized treatment, unlike the treatment of symptoms that has been common practice, is based on evidence that an individual's genotype (genetic bar code). For example, heart disease can be caused either by a mutation in certain genes, or by environmental factors such as diet or exercise. Doctors can easily diagnose a person with heart disease once they have symptoms. However, doctors can't easily tell what the cause for the heart disease is in each person. Thus, all people receive the same treatment regardless of underlying cause of the disease. In the future, a panel of genetic tests for heart disease might reveal the specific genetic factors that are involved in a given person. People with a specific mutation may be able to receive treatment that is targeted to that mutation, thereby treating the cause of the disease rather than just the symptoms.

Genetic Chips

Theoretically everything that could be known about us genetically is known (Chan and Chia 2003). Today information containing the function of our kidneys or brain, heart disease, manifestation of whole range of cancers and even ability to cope with stress or our proneness to depression are available. It is only a matter of reading out and then analyzing our genetic blue prints. It can be employed to enhance people's understanding of themselves and the world.

Check Your Progress II

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

1) What is the significance of Dolly?

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2) What are some of the future prospects offered by genetic engineering?

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2.5 CLONING AND GENETIC MANIPULATION

Cloning and genetic manipulation works towards ever increasing precision of control over what we are biologically as human beings. In 1997, a sheep named Dolly was supposedly cloned, making her the first "artificial" mammal on Earth (Wilmut, Ian and Highfield 2006).

It has been speculated that the cloning of humans could be employed to create an unstoppable "designer army". This army would be made up of genetically identical, enhanced individuals with great physical strength and artificial immunity to many diseases. The members of this hypothetical army would most likely be capable of enduring extreme physical hardship, and could even be "tailored" to the environment in which they were fighting. For example, an army sent to the tropics would be immune to tropical diseases and would be capable of fighting in extremely hot weather. An army sent to the Arctic Circle would be tolerant of exposure to very cold temperatures for long period of time.

There is a possibility of taking human DNA and clone specific organs or body parts, such as the liver, spine, or skin. This type of cloning cannot be seen as anything but a good thing. It could save thousands of lives by replacing damaged organs or body parts - for example, cloning skin for a burn victim, a spine for a paraplegic, or a liver for a hepatitis victim. This technology could vastly reduce the demand for donated organs while simultaneously eliminating the possibility of organ trade and organ rejection among patients. In addition, it would eliminate the need for immunosuppressive drugs that render organ recipients vulnerable to common diseases.

2.6 GENETIC ENGINEERING

Genetic engineering is a radical new technology, one that breaks down fundamental genetic barriers-not only between species, but also between humans, animals, and plants (Cummins 2010).It is the term applied to techniques that alter the genes (hereditary material) or combination of genes in an organism. The cells of all living organisms contain genes. Genes carry chemical information that determines the organism's characteristics. In this way scientists can give the organism and its descendents different traits. By combining the genes of dissimilar and unrelated species, permanently altering their genetic codes, novel organisms are created that will pass the genetic changes onto their offspring through heredity. Beginning in the 1970's, scientists developed ways to introduce individual genes into cells or into plants, animals, or other organisms. Scientists are now snipping, inserting, recombining, rearranging, editing, and programming genetic material. Animal genes and even human genes are being inserted into plants or animals creating unimagined transgenic life forms. For the first time in history, human beings are becoming the architects of life. Bio-engineers will be creating tens of thousands of novel organisms over the next few years.

2.7 HUMAN GENETIC ENGINEERING

Human genetic engineering is about genetically engineering human beings by modifying their genotypes before birth. The Genotype is the genetic constitution of an individual with respect to a particular character under consideration. This is done to control the traits possessed by the individual after his/her birth. The cells of our body contain encoded information about the body's growth, structure and functioning in the form of genes. Human genetic engineering aims at decoding this information and applying it to the welfare of mankind.

There are two types of genetic engineering. They are: **Somatic modification:** Genes are added to the cells. This can prove to be a cure for diseases caused by defective genes. Somatic modifications cannot be inherited. **Germline modification:** In this form of human genetic engineering, genes in the early embryos are changed. The genes modified in this way are inheritable. This is an effective form of Genetic engineering, as it results in permanent modifications.

Human genetic engineering can be classified as positive genetic engineering and negative genetic engineering. In the positive type of genetic engineering, the positive traits of individuals are enhanced. This can mean increasing longevity or increasing human capacity. The negative genetic engineering is about introducing the good copy of a certain gene into the cells of a living being. Consequently, the suffering characteristic to genetic diseases can be reduced to a great extent.

Advantages

Gene therapy is one of the most important benefits of human genetic engineering. Over the past decade, gene therapy has succeeded in finding treatments for certain heart diseases. Researchers hope to find cures for all the genetic diseases. This will result in a healthier and more evolved human race. A future benefit of human genetic engineering is that a fetus with a genetic disorder will be treated before the baby is born. Parents will be able to look forward to a healthy baby. In case of in-vitro fertilization, gene therapy can be used for embryos before they are implanted into the mother. Genes can be cloned to produce pharmaceutical products of superior quality. Researchers are hopeful about being able to bio-engineer plants or fruits to contain certain drugs.

Disadvantages

The process of cloning can lead to risking the fundamental factors such as the individuality and the diversity of human beings. Ironically, man will become just another man-made thing (Song 2002). There are certain social aspects to human genetic engineering. This new form of medical treatment can impose a heavy financial burden on the society. Along with its feasibility, its affordability will also determine its popularity. Though it seems easy to cure diseases by genetic modifications, gene therapy may manifest side effects. While treating one defect, it may cause another. Any given cell is responsible for many activities and manipulating its genes may not be that easy. Human genetic engineering is a widely growing field. It can work miracles. But its benefits and threats need to be assessed carefully. The potential advantages of the field can come into reality only if the Genetic engineering of humans is handled with responsibility.

2.8 STEM CELL RESEARCH

We have seen in the first part how hereditary play a very important role in our life. It discussed and summarized the processes of inheritance and this chapter would reveal how changes to genes at the molecular level influence large-scale metabolic and physical Characteristics. It also discussed the organization and internal structure of cells. This section also deals with stem cells. Stem cells have the remarkable potential to develop into many different cell types in the body during early life and growth. In addition, in many tissues they serve as a sort of internal repair system, dividing essentially without limit to replenish other cells as long as the person or animal

is still alive. When a stem cell divides, each new cell has the potential either to remain a stem cell or become another type of cell with a more specialized function, such as a muscle cell, a red blood cell, or a brain cell.

Stem cells can be defined as:-

Self-renewal: the ability to divide itself into exact copies numerous times, without changing into specific cell types. **Potency:** the ability to divide itself into cells that will form specific cell types that will build special tissues in the body (heart, brains, blood). Embryos formed during the blastocyst phase of embryological development (embryonic stem cells) and adult tissue (adult stem cells). Both types are generally characterized by their potency, or potential to differentiate into different cell types (such as skin, muscle, bone, etc.). Stem cells are distinguished from other cell types by two important characteristics. First, they are unspecialized cells capable of renewing themselves through cell division, sometimes after long periods of inactivity. Second, under certain physiologic or experimental conditions, they can be induced to become tissue- or organ-specific cells with special functions. In some organs, such as the gut and bone marrow, stem cells regularly divide to repair and replace worn out or damaged tissues. In other organs, however, such as the pancreas and the heart, stem cells only divide under special conditions (Humber and Almedev 2006).

Until recently, scientists primarily worked with two kinds of stem cells from animals and humans: embryonic stem cells and non-embryonic somatic or adult stem cells. The functions and characteristics of these cells will be explained in this document. Scientists discovered ways to derive embryonic stem cells from early mouse embryos nearly 30 years ago, in 1981. The detailed study of the biology of mouse stem cells led to the discovery, in 1998, of a method to derive stem cells from human embryos and grow the cells in the laboratory. These cells are called human embryonic stem cells. The embryos used in these studies were created for reproductive purposes through in vitro fertilization procedures. When they were no longer needed for that purpose, they were donated for research with the informed consent of the donor. In 2006, researchers made another breakthrough by identifying conditions that would allow some specialized adult cells to be "reprogrammed" genetically to assume a stem cell-like state. This new type of stem cell is called Induced pluripotent stem cells (IPSC's).

Stem cells are important for living organisms for many reasons. In the 3- to 5-day-old embryo, called a blastocyst, the inner cells give rise to the entire body of the organism, including all of the many specialized cell types and organs such as the heart, lung, skin, sperm, eggs and other tissues. In some adult tissues, such as bone marrow, muscle, and brain, discrete populations of adult stem cells generate replacements for cells that are lost through normal wear and tear, injury, or disease. Given their unique regenerative abilities, stem cells offer new potentials for treating diseases such as diabetes, and heart disease. However, much work remains to be done in the laboratory and the clinic to understand how to use these cells for cell based therapies to treat disease, which is also referred to as regenerative or reparative medicine. Laboratory studies of stem cells enable scientists to learn about the cells' essential properties and what makes them different from specialized cell types. Scientists are already using stem cells in the laboratory to screen new drugs and to develop model systems to study normal growth and identify the causes of birth defects.

2.9 SOURCES OF STEM CELL

Research on stem cell continues to advance knowledge about how an organism develops from a single cell and how healthy cells replace damaged cells in adult organisms. Stem cell research is one of the most fascinating areas of contemporary biology, but, as with many expanding fields of scientific inquiry, research on stem cells raises scientific questions as rapidly as it generates new discoveries.

EMBRYONIC STEM CELLS

Embryonic Stem Cells as their name suggest, are derived from embryos. Most embryonic stem cells are derived from embryos that develop from eggs that have been fertilized in vitro -in an in vitro fertilization [javascript:glosspop\('ivf'\)](#) clinic—and then donated for research purposes with informed consent of the donors. They are not derived from eggs fertilized in a woman's body. Embryonic stem cells are derived from a four- or five-day-old human embryo that is in the blastocyst phase of development. The embryos are usually extras that have been created in IVF (in vitro fertilization) clinics where several eggs are fertilized in a test tube, but only one is implanted into a woman (Humber and Almedev 2006).

Sexual reproduction begins when a male sperm fertilizes a female ovum (egg) to form a single cell called a zygote. The single zygote cell then begins a series of divisions, forming 2, 4, 8, 16 cells, etc. After four to six days - before implantation in the uterus - this mass of cells is called a blastocyst. The blastocyst consists of an inner cell mass (embryoblast) and an outer cell mass (trophoblast). The outer cell mass becomes part of the placenta, and the inner cell mass is the group of cells that will differentiate to become all the structures of an adult organism. This latter mass is the source of embryonic stem cells - totipotent cells (cells with total potential to develop into any cell in the body).

ADULT STEM CELLS

An adult stem cell is thought to be an undifferentiated cell, found among differentiated cells in a tissue or organ that can renew it and can differentiate to yield some or all of the major specialized cell types of the tissue or organ. The primary roles of Adult Stem Cells in a living organism are to maintain and repair the tissues in which they are found. Scientists also use the term Somatic Stem Cells instead of adult stem cells, where somatic refers to cells of the body (not the germ cells, sperm or eggs) (Humber and Robert F. Almedev 2006). Unlike embryonic stem cells, which are defined by their origin, the origin of adult stem cells in some mature tissues is still under investigation.

Adult stem cells have been identified in many organs and tissues, including brain, bone marrow, peripheral blood, blood vessels, skeletal muscle, skin, teeth, heart, gut, liver, ovarian epithelium, and testis. They are thought to reside in a specific area of each tissue (called a "stem cell niche"). In many tissues, current evidence suggests that some types of stem cells are pericytes, cells that compose the outermost layer of small blood vessels. Stem cells may remain quiescent (non-dividing) for long periods of time until they are activated by a normal need for more cells to maintain tissues, or by disease or tissue injury.

Typically, there is a very small number of stem cells in each tissue, and once removed from the body, their capacity to divide is limited, making generation of large quantities of stem cells difficult. Scientists in many laboratories are trying to find better ways to grow large quantities of adult stem cells in cell culture and to manipulate them to generate specific cell types so they can be used to treat injury or disease. Some examples of potential treatments include regenerating bone using cells derived from bone marrow, developing insulin-producing cells for type 1 diabetes, and repairing damaged heart muscle following a heart attack with cardiac muscle cells.

INDUCED PLURIPOTENT STEM CELLS (IPSC's)

Induced pluripotent stem cells (IPSC's) are adult cells that have been genetically reprogrammed to an embryonic stem cell-like state by being forced to express genes and factors important for maintaining the defining properties of embryonic stem cells. Although these cells meet the defining criteria for pluripotent stem cells, it is not known if IPSC's and embryonic stem cells differ in clinically significant ways. Mouse IPSC's were first reported in 2006, and human IPSC's were first reported in late 2007. Mouse IPSC's demonstrate important characteristics of pluripotent stem cells, including expressing stem cell markers, forming tumors containing cells from all three germ layers, and being able to contribute to many different tissues when injected into mouse embryos at a very early stage in development. Human IPSC's also express stem cell markers and are capable of generating cells characteristic of all three germ layers.

Although additional research is needed, IPSC's are already useful tools for drug development and modeling of diseases, and scientists hope to use them in transplantation medicine. Viruses are currently used to introduce the reprogramming factors into adult cells, and this process must be carefully controlled and tested before the technique can lead to useful treatments for humans. In animal studies, the virus used to introduce the stem cell factors sometimes cause cancers. Researchers are currently investigating non-viral delivery strategies. In any case, this breakthrough discovery has created a powerful new way to "de-differentiate" cells whose developmental fates had been previously assumed to be determined. In addition, tissues derived from IPSC's will be a nearly identical match to the cell donor and thus probably avoid rejection by the immune system. The IPSC strategy creates pluripotent stem cells that, together with studies of other types of pluripotent stem cells, will help researchers learn how to reprogram cells to repair damaged tissues in the human body.

2.10 POTENCY AND PROPERTIES OF STEM-CELLS

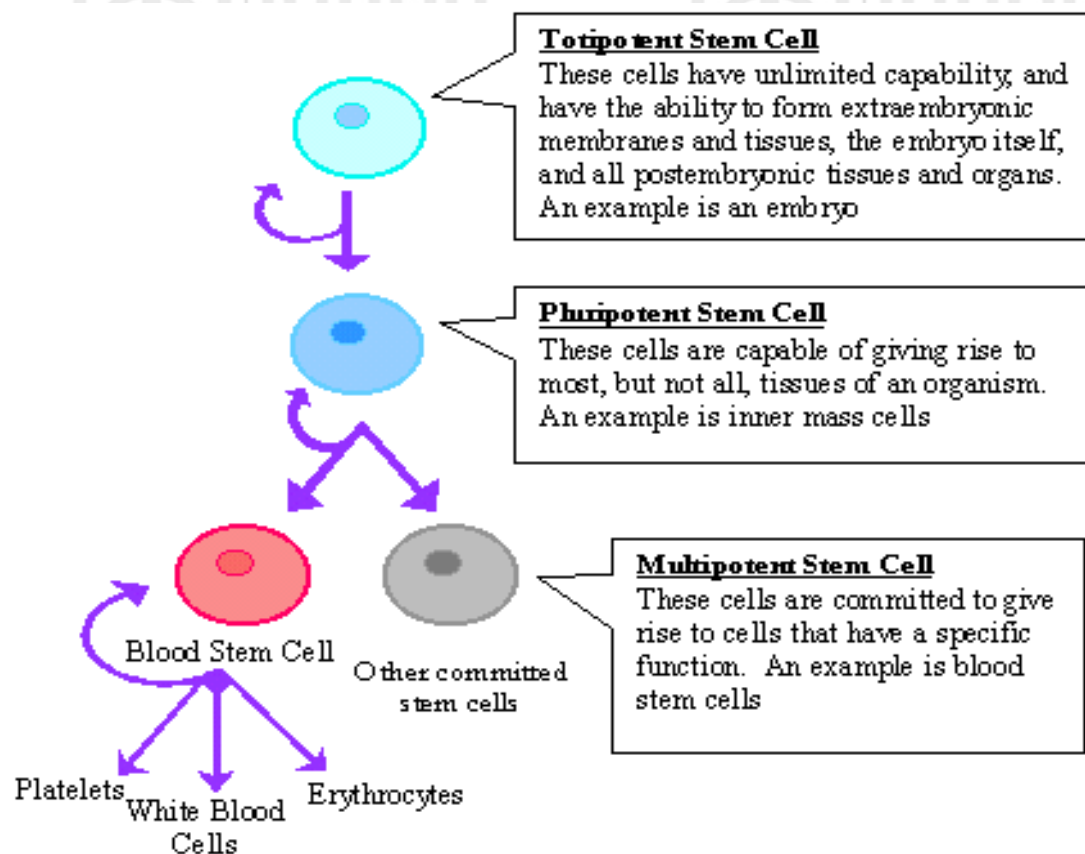
Stem cells are categorized by their potential to differentiate into other types of cells. Embryonic stem cells are the most potent since they must become every type of cell in the body. The full classification includes:

Totipotent - the ability to differentiate into all possible cell types. Examples are the zygote formed after the fertilization and the first few cells that result from the division of the zygote.

Pluripotent - the ability to differentiate into almost all cell types. Examples include embryonic stem cells and cells that are derived from the mesoderm, endoderm, and ectoderm germ layers that are formed in the beginning stages of embryonic stem cell differentiation.

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of stem cell research is politically charged, prompting biologists to begin engaging in ethical debates, and generating in the general public an unusually high level of interest in this aspect of biology. but excitement notwithstanding, there is a long way to go in basic research before new therapies will be established, and now the pressure is on for scientists and clinicians to deliver.

Check Your Progress III

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

1) What are some of the advantages of Genetic Engineering?

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2) What is a pluripotent cell?

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2.11 LET US SUM UP

One of the characteristic features of human being is that he/ she can go beyond many limits and move to higher levels of existence. This quality of going beyond or being superior to was manifested through technology especially in Genetic science. We must not be the slaves of the technology. Techniques, however grand they may be, must reduce man to technical animal, the king of the slaves of technique. While creating and relying on the Human- made environment, we ourselves is being modified with new outlook and orientations. The technology limits our outlook in certain way leading to the consumer outlook, materialistic and military orientations etc. The human being now becomes different and he/she can be called the technological man/woman because of the dominance of technology over his/her life. From a contextual study we become aware of the very nature of human being and how we are constantly becoming different through the advancement in Genetics and other technological developments. So in this unit we have studied the basics of Genetic engineering and Stem-cell research.

2.11 KEY WORDS

Genetic Engineering: The deliberate modification of the characteristics of an organism by manipulating its genetic material.

Germline: Genetic material in a cell lineage that is passed down through the gametes before it is modified by somatic recombination or maturation.

Stem cell: An undifferentiated cell of a multicellular organism that is capable of giving rise to indefinitely more cells of the same type, and from which certain other kinds of cell arise by differentiation.

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UNIT 3 BASICS OF HUMAN GENOME PROJECT

Contents

- 3.0 Objectives
- 3.1 Introduction
- 3.2 History of HGP
- 3.3 Human Genome Project: An Overview
- 3.4 Goals of HGP
- 3.5 Advantages of Human Genome Project
- 3.6 Achievement of Human Genome Project
- 3.7 HGP: Future Prospects
- 3.8 Philosophical Reflections
- 3.9 Let Us Sum Up
- 3.10 Key Words
- 3.11 Further Readings and References

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 main 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 Φ -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 Wellcome 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?

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

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 unsequenced.

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?

.....

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 defects 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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UNIT 4 ETHICAL, LEGAL AND SOCIAL ISSUES

Contents

- 4.0 Objectives
 - 4.1 Introduction
 - 4.2 Elsi Research Goals
 - 4.3 Ethical Issues
 - 4.4 Legal Issues
 - 4.5 Social Issues
 - 4.6 Critical Remarks
 - 4.7 Some Large Philosophical Issues
 - 4.8 Let us Sum Up
 - 4.9 Key Words
 - 4.10 Further Readings and References
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4.0 OBJECTIVES

- To give the students the general overview of ELSI programs and its significance to genetic engineering.
 - To see the significance of ELSI, particularly in genetic research.
 - To have a comprehensive notion of both the positive and negative side effects and consequences of HGP.
 - To give an overview of the ethical, legal and social issues related to genetic research.
 - To see some of the dangers posed and possibilities offered by HGP and such cutting-edge technologies
-

4.1 INTRODUCTION

Rapid advances in the science of genetics and its applications have presented new and complex ethical and policy issues for individuals and society. Ethical, Legal and Social Issues (ELSI) programs that identify and address these implications have been an integral part of the U.S. Human Genome Project (HGP) since its inception. These programs have resulted in a body of work that promotes education and helps guide the conduct of genetic research and the development of related medical and public policies.

A continuing challenge is to safe guard the privacy of individuals and groups who contribute DNA samples for large-scale sequence-variation studies. Other concerns have been to anticipate how the resulting data may affect concepts of race and ethnicity; identify potential uses (or misuses) of genetic data in workplaces, schools, and courts; identify commercial uses; and foresee impacts of genetic advances on the concepts of humanity and personal responsibility. The project's goals included not only identifying all of the approximately 24,000 genes in the human genome, but also to address the ethical, legal, and social issues (ELSI) that might arise from the availability of genetic information. Five percent of the annual budget was allocated to address the ELSI arising from the project.

The main criticism of ELSI is the failure to address the conditions raised by population-based research, especially with regard to unique processes for group decision-making and cultural worldviews. Genetic variation research such as HGP is group population research, but most ethical guidelines, according to Harry, focus on individual rights instead of group rights. She says the research represents a clash of culture: indigenous people's life revolves around collectively and group decision making whereas the Western culture promotes individuality. Harry suggests that one of the challenges of ethical research is to include respect for collective review and decision making, while also upholding the Western model of individual rights.

4.2 ELSI RESEARCH GOALS

The HGP is probably one of the most profound research efforts in human history, which will certainly impact all of us because of the many implications for medicine and human health. The Genetic advancements and Human Genome Project has the potential to heal and restore the brokenness of creation. However it has serious ethical, social, philosophical and religious implications. Ethical, legal and social implications of the Human Genome Project are privacy and confidentiality, psychological impact and stigmatization, genetic testing, reproductive issues, education, standards, and quality control, commercialization, conceptual and philosophical implications.

1. **Fairness and privacy:** fairness in the use of genetic information. Who should have access to your genetic information? (Chan and Chia 2003)
2. **Psychological stigmatization:** how does knowing your predisposition to disease affect an individual?
3. **Genetic testing:** should screening be done when there is no treatment available?
4. **Reproductive issues:** Reproductive issues including adequate informed consent for complex and potentially controversial procedures, use of genetic information in reproductive decision making, and reproductive rights.
5. **Clinical issues:** implementation of standards and quality control measures in testing procedures.
6. **Commercialization:** commercialization of products includes property rights (patents, copyrights, and trade secrets) and accessibility of data and materials.

Some of the other related issues that are related to ELSI are:

- Examine issues surrounding the completion of the human DNA sequence and the study of human genetic variation.
- Examine issues raised by the integration of genetic technologies and information into health care and public health activities.
- Examine issues raised by the integration of knowledge about genomics and gene-environment interactions in non-clinical settings.
- Explore how new genetic knowledge may interact with a variety of philosophical, theological, and ethical perspectives.
- Explore how racial, ethnic, and socioeconomic factors affect the use, understanding, and interpretation of genetic information; the use of genetic services; and the development of policy.

Check Your Progress I

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

1) What is ELSI and give its significance?

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2) What are some of the research goals of ELSI?

.....

4.3 ETHICAL ISSUES

It is often asked whether it is right for human beings to manipulate human genes at all, as if this in some sense “playing God” by altering fundamental aspects of human makeup which are God’s prerogative only, or simply that it is a dangerous “tampering with nature” in a way which we have neither the right nor the skill to do. We need to worry about whether genetic technology generated from the HGP will make us less accepting of people who are different. For example, if it is possible to predict and prevent the birth of a child with a gene-related disorder, how will we react to children who have that disorder? At the same time the non reproductive cloning requires abortion (Kilner, Pentz and Young 1997). Here life is created specifically for the purpose of destroying it and then cannibalizing it essentially for spare parts. Human life is intrinsically sacred even at this early stage of development.

Genetic modification of trees to reduce Co2 levels is a new attempt in genetic world. It aims to bring a benefit to all people which would be treating them as an end in themselves. A comparison with the duty based ethics of Kant would show more light into this. Immanuel Kant says that we should treat each person as an end in themselves and not merely as a means to an end. Kant see it as a duty to implement this technology, provides we look as much care as possible to ensure that it was for the welfare (Lewis 2007). Thus the humanity will benefit from a stabilization of the climate by genetically modified trees. It is Kantian duty to act in the interests of future generations to treat them all as ends in themselves. However if something goes wrong, then humanity would suffer from changes to their eco-system.

What are some of the Ethical considerations for using gene therapy?

Some questions to consider are:

- What is normal and what is a disability or disorder, and who decides?
- Are disabilities diseases? Do they need to be cured or prevented?
- Does searching for a cure demean the lives of individuals presently affected by disabilities?
- Is somatic gene therapy (which is done in the adult cells of persons known to have the disease) more or less ethical than gremlin gene therapy (which is done in egg and sperm cells and prevents the trait from being passed on to further generations)? In cases of somatic gene therapy, the procedure may have to be repeated in future generations.

- Preliminary attempts at gene therapy are exorbitantly expensive. Who will have access to these therapies? Who will pay for their use?

What are the ethical issues surrounding gene?

Because gene therapy involves making changes to the body's set of basic instructions, it raises many unique ethical concerns. The ethical questions surrounding gene therapy include:

- How can “good” and “bad” uses of gene therapy be distinguished?
- Who decides which traits are normal and which constitute a disability or disorder?
- Will the high costs of gene therapy make it available only to the wealthy?
- Could the widespread use of gene therapy make society less accepting of people who are different?
- Should people be allowed to use gene therapy to enhance basic human traits such as height, intelligence, or athletic ability?
- Short-lived nature of gene therapy – Before gene therapy can become a permanent cure for any condition, the therapeutic DNA introduced into target cells must remain functional and the cells containing the therapeutic DNA must be long-lived and stable. Problems with integrating therapeutic DNA into the genome and the rapidly dividing nature of many cells prevent gene therapy from achieving any long-term benefits. Patients will have to undergo multiple rounds of gene therapy.
- Immune response – Anytime a foreign object is introduced into human tissues, the immune system has evolved to attack the invader. The risk of stimulating the immune system in a way that reduces gene therapy effectiveness is always a possibility. Furthermore, the immune-system's enhanced response to invaders that it has seen before makes it difficult for gene therapy to be repeated in patients.
- Problems with viral vectors – Viruses, the carrier of choice in most gene therapy studies, present a variety of potential problems to the patient —toxicity, immune and inflammatory responses, and gene control and targeting issues. In addition, there is always the fear that the viral vector, once inside the patient, may recover its ability to cause disease.
- Multi-gene disorders – Conditions or disorders that arise from mutations in a single gene are the best candidates for gene therapy. Unfortunately, some of the most commonly occurring disorders, such as heart disease, high blood pressure, Alzheimer's disease, arthritis, and diabetes, are caused by the combined effects of variations in many genes. Multi-gene or multi-factorial disorders such as these would be especially difficult to treat effectively using gene therapy.

Current gene therapy research has focused on treating individuals by targeting the therapy to body cells such as bone marrow or blood cells. This type of gene therapy cannot be passed on to a person's children. Gene therapy could be targeted to egg and sperm cells (germ cells), however, which would allow the inserted gene to be passed, on to future generations. This approach is known as germ-line gene therapy (Harwood 1994).

The idea of germ-line gene therapy is controversial. While it could spare future generations in a family from having a particular genetic disorder, it might affect the development of a fetus in

unexpected ways or have long-term side effects that are not yet known. Because people who would be affected by germ-line gene therapy are not yet born, they can't choose whether to have the treatment (Harwood 1994). Because of these ethical concerns, the U.S. Government does not allow federal funds to be used for research on germ-line gene therapy in people.

- At the ethical level we also need to ask the following questions:
- What is normal and what is a disability or disorder, and who decides?
Are disabilities diseases?
- Do they need to be always cured or prevented?
- Does searching for a cure demean the lives of individuals presently affected by disabilities?
- Is somatic gene therapy (which is done in the adult cells of persons known to have the disease) more or less ethical than germ line gene therapy (which is done in egg and sperm cells and prevents the trait from being passed on to further generations)?
- In cases of somatic gene therapy, the procedure may have to be repeated in future generations. Can we give this burden to the future generation?

Preliminary attempts at gene therapy are exorbitantly expensive. Who will have access to these therapies? Who will pay for their use? The eradication of disease through germ-line therapy might not seem, by itself, to raise many ethical questions. After all, humans have eradicated the smallpox virus from the world, why not diseases with genetic components? Do doctors not have the moral obligation to provide the very best treatment to their patients and would not the eradication of the disease be more cost effective in the long run than continually treating adults with somatic gene therapy? The main ethical problem arises in defining a "treatable" disease. Some might say that eradication of a genetic disease for which there no treatment is and which is always fatal, should be pursued with all means possible. Others say that this would be the start of a slippery slope moving on toward the treatment of less obvious diseases and then to genetic enhancement.

Some argue that if the technology is advanced in order to eradicate some diseases, it will inevitably be used by parents wishing to "enhance" their children, giving them the genes for raven black hair and blue eyes or athletic prowess. It was serious ethical concerns about genetic enhancement that prompted the Council of Europe to adopt the Convention for the Protection of Human Rights and Dignity of the Human Being with Regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine. Article 13 of the Convention states that "an intervention seeking to modify the human genome may only be undertaken for preventive, diagnostic or therapeutic purposes and only if its aim is not to introduce any modification in the genome of any descendants.

"Article 11 of the UNESCO (Universal Declaration on the Human Genome and Human Rights) states that "practices which are contrary to human dignity, such as reproductive cloning of human beings, shall not be permitted." It is left to individual states; however, to define exactly what they believe these practices to be. Thus, while some countries, such as the signatories to the European Convention, may prohibit germ-line therapy, others may not. It is the existence of national differences in regulation of research on human embryos that has allowed controversial

research to be performed, for example, in Singapore. Regulation has thus slowed down the progress of research but not prevented it.

Another ethical consideration with respect to germ-line therapy is defining what is normal, what a disability is, and what a disease is. Which of the genetic variations within a population ought to be eradicated, if any? In trying to eradicate a certain variation, are we demeaning those in the population who currently carry the gene? Somatic gene therapy has its own, less controversial, set of ELS implications. These may be less ominous than eugenics but are of perhaps more immediate concern, given the more advanced state of the technology. Effectively, gene therapy involves the introduction of a properly functioning gene into target tissues in the hopes that it will be translated into a properly functioning protein, which will mask the malfunctioning protein. Often the new gene is placed into a modified virus, which is then introduced into a patient in the hope that the gene will be introduced into a tissue and properly expressed.

Such types of therapy, after much research on laboratory in animals, have now reached the clinical trial stage. Unfortunately, what works for a mouse does not always work for a human being. In one highly publicized case, a patient, Jesse Gelsinger, was given an injection of a virus in the hope of introducing a protein into the liver. Mouse studies showed good absorption of the gene into the liver; however, the mouse has a much higher concentration of viral receptors on its liver cells than do humans. The virus did not absorb well into the human patient and, for still unknown reasons, created a massive immune response, causing the patient to die. The original plan for the trials had been to use the virus only on children in a coma caused by the lack of the particular liver enzyme; however, ethical and safety reviews caused the researchers to change the trial direction and use adults only.

Many questions are now being asked regarding the ethics and scientific judgment of those performing such clinical trials. How well are "volunteer" patients informed of the possible risks and benefits? How objective are investigators who have equity in the companies that are funding the trials? One of the risks at this stage of gene therapy is the excessive public anticipation, created in part by some researchers, with respect to future benefits. This anticipation may turn to public distrust of science, if the benefits fail to be realized and problems such as that in the Gelsinger case continue to occur. Some clinical trials have shown positive results, and so there is still hope that somatic gene therapy will become a powerful medical tool (Green 2007).

4.4 LEGAL ISSUES

Medical therapies: Scientists are now discovering the genes which "trigger" various genetic diseases which, in turn, constitute a large part of the inherited causes of the suffering of humanity. For example, the genes which express Huntington's disease, a serious affliction, have been identified on the human genome. Their discovery permits the conduct of extremely accurate tests which can now identify those people who carry and may transmit this genetic condition. That knowledge would, theoretically, in combination with prenatal tests and abortion, permit the future elimination of carriers of Huntington's. So we need to ask the following questions:

- Is such elimination of carriers of Huntington's desirable? Can it be distinguished from the abortion of a foetus with Down syndrome?
- Where does this process of medical elimination of the results of "defective" genes begin and end?

- Is there a less life-destructive means of using the genetic information to delay the onset or diminish the symptoms of Huntington's disease whilst respecting the life of a person born with those genes or others like it?

Criminal Law: For the lawyer, the discovery of genetic causes of disorders and of some antisocial conduct may have implications for the future. The criminal law is built upon a general hypothesis of free will. For the crime to be established it is normally necessary to prove both the act of the accused and the will occasioning that act. Some related issues are:

- What are the implications for the law of discovering that, in some cases at least, for some people, the act is practically nothing but the product of a genetic characteristic?
- Can we persist, in all cases, with the unquestioned hypothesis of free will in the face of scientific knowledge which casts doubt upon it?
- Privacy and Confidentiality: The basic rule of the healthcare professions has long been respect for the confidences of the patient. This rule goes back at least to the Hippocratic Oath. It existed in ancient civilisations.
- When a disorder is of a genetic characteristic, is the "patient" the individual or the entire family?
- Does a family in such circumstances have a right to override the wishes of the patient and to secure data about the patient's genes relevant to genetic features important for them all?
- Should a patient have a right not to know the determinants of his or her future medical conditions?

Third Party Interests: This last question leads to the rights of third parties. Should an employer have a right to require an employee to submit to genetic testing to show, with greater perfection, the likely future health status of the employee? Should an insurer be entitled to secure a detailed genetic profile of the insured? Until now, insurance has generally involved the sharing, within the community, of the risks attached to medical conditions which are largely unpredictable.

If such conditions can be predicted with perfect or near perfect accuracy, would that not shift the scales unfairly to the advantage of insurers? Where insurers can require those seeking insurance to submit to old-fashioned medical tests, is it sensible to close off knowledge of the best medical information that may be made available by genetic tests?

Intellectual Property: One of the key issues of genetic research concerns the desirability of permitting the patenting of human genes or their sequences as the basis for future therapeutic applications. Of course, in every country, the patentability of such matter depends upon the terms of the local law on intellectual property protection. That law is itself normally the product of national legislation and is often influenced by international law. At conferences on the genome, strong views are commonly expressed by participants from developing countries and elsewhere about this topic. They urge that the human genome is the common heritage of humanity. That it belongs to the human species as a whole - some say to God - and not to private corporations engaged in research, however potentially beneficial such research may prove to be. They point to the fact that Watson and Crick never attempted to secure the slightest commercial advantage for themselves from their discoveries.

Human Rights: An important element in UNESCO's Universal Declaration on Human rights and the Human Genome, to which I will now turn, is the attempt to reconcile the development of

genetic technology and research on the human genome with fundamental human rights and human dignity inhering in every individual (Podimattom 2003).

4.5 SOCIAL ISSUES

The HGP is rich with promises, but also fraught with social implications. It is quite likely that the new knowledge from the HGP will be used in ways that don't always have to do with health. Our challenge is to reduce any negative impacts, which result from the misuse of genetic information. Researchers, scientists, business and governmental people who are involved in the HGP must avoid any activity that could cause harm while they pursue professional and personal goals.

On the other hand, individuals not directly participating in genetic research are responsible to educate themselves and seek information about potential risks and benefits of genetic research and about the interventions that it produces before they utilize new genetic technologies. Individuals with access to information about their genetic endowments will be able to predict their susceptibility to genetically related disorders in the future (Chan and Chia 2003). Threats evolve around the major question of whether or not an individual has access to genetic technologies. One type of threat occurs when individuals are denied access to technologies that would benefit them. The likelihood of genetic technologies becoming available to some peoples but not to others, and that a major determinant of access will be wealth, raises profound social issues. In view of Global collaboration the HGP should also has to deal with rules especially fair regulations that concern the access and uses of the new information.

The regulation of access of technologies and information obtained from the HGP should be handled very carefully and is probably one of the most difficult tasks for the future. This is especially true since the project has international dimensions and opinions on this issue differ among participating countries. That leads into another question, whether countries that do not directly contribute to the success of the HGP should be treated differently in terms of access to information and technologies being generated from the HGP.

Humans are the moral agents in this world with a capacity to think, evaluate, choose, communicate and articulate. It has been argued that the most significant issue genetic science forces on society concerns the understanding of human nature. Objectification also represents a fundamental breach of human dignity. To treat persons who are the sources of genetic material for cloning or persons who are created through cloning as mere objects, means or instruments violates the religious principle of human dignity as well as the secular principle of respect for persons.

Genetic Discrimination

One of the problems some fear might result from knowledge of the human genome is the emergence of a whole population of socially marginalized individuals, unable to obtain a job, a family, insurance, or health care and stigmatized by the rest of society. Insurance companies already insist that those identified at risk of Huntington's disease must take a genetic test. If the results are positive, insurance is frequently refused. Insurance companies are on record as saying that if genetic information was available, they would use it in their risk assessment.

In Canada, the refusal to insure a Huntington's patient does not have dire consequences; in general, public insurance covers many aspects of care, though the level of care varies across the country and the coverage for pharmaceuticals is less clear. In countries without a public health insurance system, however, the plight of such a non-insured person can be a nightmare. Care may be available but finding it is very difficult. As more genetic tests become available, insurance is likely to be more and more expensive for those carrying what the insurance companies deem to be risky genes.

The public insurance schemes may also start to feel the pressure for such genetic testing, and be forced to make policy decisions based on the funding available and the knowledge of genetic predisposition to disease within populations. Gene therapy is at the experimental stage at this point but will certainly be very expensive when it first comes into regular use. Who will pay for it? If not public insurance, will the therapy be available only to rich people, thus creating an ever widening gap between groups in society, based on both money and genetic inheritance? Employers may also want access to genetic information. Some genes might reveal a susceptibility to environmental damage that was incompatible with a certain workplace environment. Employers might choose to screen out workers carrying that gene rather than trying to improve the environment. Individuals with genes associated with certain behavioural traits might also be excluded from the workplace (Olyshevsky 2002).

Racial Discrimination

Although no genetic-employment discrimination case has been brought before U.S. federal or state courts, in 2001 the Equal Employment Opportunity Commission (EEOC) settled the first lawsuit alleging this type of discrimination. EEOC filed a suit against the Burlington Northern Santa Fe (BNSF) Railroad for secretly testing its employees for a rare genetic condition that causes carpal tunnel syndrome as one of its many symptoms. BNSF claimed that the testing was a way of determining whether the high incidence of repetitive-stress injuries among its employees was work-related. Besides testing for this rare problem, company-paid doctors also were instructed to screen for several other medical conditions such as diabetes and alcoholism. BNSF employees examined by company doctors were not told that they were being genetically tested. One employee who refused testing was threatened with possible termination.

On behalf of BNSF employees, EEOC argued that the tests were unlawful under the Americans with Disabilities Act because they were not job-related, and any condition of employment based on such tests would be cause for illegal discrimination based on disability. The lawsuit was settled quickly with BNSF agreeing to everything sought by EEOC. Besides the BNSF case, the Council for Responsible Genetics claims that hundreds of genetic-discrimination cases have been documented. In one case, genetic testing indicated that a young boy had Fragile X Syndrome, an inherited form of mental retardation. The insurance company for the boy's family dropped his health coverage, claiming the syndrome was a pre-existing condition. In another case, a social worker lost her job within a week of mentioning that her mother had died of Huntington's disease and that she had a 50% chance of developing it.

Despite claims of hundreds of genetic-discrimination incidents, an article from the January 2003 issue of the European Journal of Human Genetics reports a real need for a comprehensive

investigation of these claims. The article warns that many studies rely on unverified, subjective accounts from individuals who believe they have been unfairly subjected to genetic discrimination by employers or insurance companies. Rarely are these subjective accounts assessed objectively to determine whether actions taken by employers and insurers were truly based on genetic factors or other legitimate concerns.

The cultural implications of the concept of gentrification come into full view when they search for genes that may for example explain alcoholism, homosexuality, aggressive behaviour, or difficulties in learning are the cases in point. While in the past certain types of individual behaviour were interpreted as representations of individual life choices within the parameters of a given society, Genetification of Life interprets these same choices as ultimately constituted at the genetic level and expression at the biological level “beyond freedom and dignity” (Nelson 1994).

Check Your Progress II

Note: Use the space provided for your answers.

1) How do you related the need for privacy and genetic determination?

.....

2) Briefly describe some of the social issues connected with Human Genome Project?

.....

.....

4.6 CRITICAL REMARKS

The Human Genome Project also include work on identifying and addressing the ethical, legal, and social issues that are and will be created by this new knowledge. The societal concerns identified include:

- Fairness in the use of genetic information by insurers, employers, courts, schools, adoption agencies, and the military, among others.
- Privacy and confidentiality of genetic information.
- Psychological impact and stigmatization due to an individual's genetic differences.
- Reproductive issues, including adequate informed consent for complex and potentially controversial procedures, use of genetic information in reproductive decision making, and reproductive rights.
- Clinical issues, including the education of doctors and other health service providers, patients, and the general public in genetic capabilities, scientific limitations, and social risks; and implementation of standards and quality-control measures in testing procedures.
- Uncertainties associated with gene tests for susceptibilities and complex conditions(e.g., heart disease) linked to multiple genes and gene-environment interactions.

- Conceptual and philosophical implications regarding human responsibility, free will vs genetic determinism, aim concepts of health and disease.
- Health and environmental issues concerning genetically modified foods (GMFs) and microbes.
- Commercialization of products, including property rights (patents, copyrights, and trade secrets) and accessibility of data and materials. Commodification of Human beings will become a major concern as human beings would turn out to be marketing products.

4.7 SOME LARGE PHILOSOPHICAL ISSUES

The new breakthroughs in genetics will question the traditional/religious understanding of the concept of God, creation and death. Are we at the beginning of a New World, where we can attain immortality? We are at the stage of Playing God for the whole Universe? Here the famous Nietzsche words would be applicable. "Is man one of God's Blunders? Or is God one of man's blunders ". Science allows us to either transcend or arrest our own aging process. "Given all the responsibility to God and to our fellow human beings, do we have a right to let experimentations and ownership of new life move ahead without public regulation?" (Evans 2002)

One of the most interesting and profound changes is not with the biotech firms or venture capitalists but with the way we see ourselves. We live in a culture that links genes or DNA sequences to complex predispositions and patterns of behavior, usually without even bothering to think about what a gene actually is. Thus when we talk about a gene for crime and gene for love as if the gene determines the behavior by rigid mechanical chain of deterministic slavery (Chan and Chia 2003). It calls us to critically analyze the human responsibility, free will vs. genetic determinism. Designer drugs like Prozac and Ritalin have the effect of modifying and muting human diversity. It has a serious impact on human diversity, dignity and human rights based on our sense of innate human nature since it is the human nature that gives us moral sense and provides us with social skills to live in a society and serves as a ground for philosophical discussions of rights, justice and morality.

Genetics advancement must not act as a barrier in our life especially in taking responsibility for our lives and the choices we make. In Germany, there is a widespread opposition to learning a lot about human genetics The Hitler Nazi regime in Germany between 1933 and 1945 killed millions of Jews, Gypsies, mental patients, and disabled people in concentration camps in the name of the pseudoscience eugenics. Any suggestion that eugenic improvements may be feasible, as a result of gained knowledge from the HGP, are alarming signs and create strong objections to the sequencing of human genome. That eugenics philosophy which lead to the horrors of National Socialism in Germany have made many people appropriately sensitive to the potential abuses of genetic science.

Genetics shortens the gap between us and other species. However it widens the view of human variation (DeGrazia 2005). There are differences among us that go to the heart of our self and our personhood. What is then considered acceptable diversity? Where is the line between medical treatment and enhancement? These are the some of the important and significant questions for the philosophers to ponder. Genetic manipulation becomes arbitrary and unjust when it reduces life to an object, when it forgets that it is dealing with a human subject. Life is the supreme good of man. First of all, anything that is harmful to it must be opposed. Only then what is good must be sought out and pursued. The question before human beings is whether they are capable of seizing the moment of creative opportunity and deciding how to make this new technology serve the well- being of human kind. If they succeed in doing so, this will go down in

history as crowning of life. On the other hand, if the human beings deal with it irresponsibly without giving their critical and reflective ability a chance, they might end up in clowning life which in no way is the choice of humanity.

4.8 LET US SUM UP

Many of the ethical, legal and social issues that are being discussed with respect to the Human Genome Project are not new. Genetic tests for a variety of diseases are currently available and some people are already struggling with the ethical and practical implications. What will change over the next few years, as a result of the Human Genome Project, is the scale of the issues and how society will have to cope with the greyer areas of genetic disease and disability. Dealing with a single gene that causes death or chronic disability is one issue; dealing with whole sets of genes whose impacts vary depending on environmental interactions is another.

The rate of scientific advancement has tended to outstrip the legislative capacity of governing bodies and there has been some media "overhype" with respect to genetic research and its potential for treatment of disease. It will be years before many of the genetic tests are available and before genetic diseases can be treated. Society as a whole must use this time to discuss and decide on how genetic information ought to be used, before the choices are made for them. It is a discussion that those with genetic dispositions to diseases such as Huntington's have long wanted to make more public.

The Human Genome Project is a remarkable breakthrough in medical science and biological study and while there are ethical questions about the use of the information, overall, knowing the map of the human genome has allowed incredible medical breakthroughs in recent years. Processes such as genetic testing and gene therapy have created more awareness about certain inherited diseases such as cystic fibrosis and sickle cell anaemia and have helped countless people alter their lifestyles so they do not fall prey to diseases such as Alzheimer's or breast cancer, which are both inherited susceptibilities. While there are still moral questions to be answered, such as those addressing the issue of altering the genome of unborn children, the Human Genome Project can certainly be identified as a benefit to the world when the data is handled by the right people? So in this unit we have taken up some of the basic ethical, legal, social and philosophical issues related to Genetic Engineering. Larger discussion and debate on these issues are really needed.

Check Your Progress III

Note: Use the space provided for your answers.

1) What is commodification of human beings?

.....

2) Describe some of the larger philosophical issues dealing with genetic engineering?

.....

4.9 KEY WORDS

Commodification: The tendency to treat humans beings as commodities or objects for use.

GM: Genetically modified (GM) foods are foods derived from genetically modified organisms. Genetically modified organisms have had specific changes introduced into their DNA by genetic engineering techniques.

GMO: A genetically modified organism (GMO) or genetically engineered organism (GEO) is an organism whose genetic material has been altered using genetic engineering techniques

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