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The Human Genome Project: An Insight to the Homo Sapiens

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ABSTRACT: We are now living in the 21st century, where we know technology has revolutionised the way of living of human life to so much so that we are now looking forward to be technologically dependent enough to do even small tasks of simple life, to complex issues of human beings. The human genome project enhances the capabilities of human beings, towards a new era where we may have possibilities of tracing even the deadliest diseases and even dealing with it. In this paper, the human genome project is being explored to make assumptions of further researches and how far the issues of healthy human society could be established is estimated. The idea is to allow the experts reflect upon the intricate progressions of science into practices of genes, and improve tools that can map genes, with the simulation of information technology. Among the main goals of the Human Genome Project (HGP) was to develop new, better and cheaper tools to identify new genes and to understand their function.

KEYWORDS: Genomes. HGP, Digital Computation, Computational analysis, unraveling the Life book, Disease predictions, fashioning the babies.

INTRODUCTION: In June 2000, the US-based National Institute of Health (NIH) and Celera Genomics Corp., a biotech company, proclaimed that they had deciphered about 90 per cent of the human genome. This decade old Human Genome (book of life) Project is trying to decipher our genes and in this process it is mixing out 12,000 letters of genetic code every minute of every day, making a list that will total more than 3 billion when finally concluded. Amazingly, more than 1,100 biologists, computer scientists and analysts at university laboratories in six different countries have been stiff at work trying to complete what some are calling biology's style of the book of life. This project is like positioning a man on the moon.

The Human Genome Project (HGP) was one of the great achievements of exploration in history - an inward voyage of encounter rather than an outward exploration of the planet or the cosmos; an international research effort to sequence and map all of the genes - together known as the genome - of members of our species, *Homo sapiens.* Accomplished in April 2003, the HGP gave us the ability, for the first time, to read nature's complete genomic blueprint for building a humanoid or anthropological being. I remember a quote from the book Quran :

"We shall show them Our portents on the horizons
And **within them- selves** until it will be
Manifest unto them that it is the Truth.
Does not your Lord suffice,
Since He is Witness over all things?"
Al-Qur'an, Surah Fusilat, 41: 53

One of these tools is genetic mapping. Genetic mapping - also called linkage mapping - can offer firm evidence that a disease transmitted from parent to child is linked to one or more genes. Mapping also provides clues about which chromosome contains the gene and precisely where the gene lies on that chromosome.

Genetic maps have been used successfully to find the gene responsible for relatively rare, single-gene inherited disorders such as cystic fibrosis and Duchenne muscular dystrophy. Genetic maps are also useful in guiding scientists to the many genes that are believed to play a role in the development of more common disorders such as heart disease, asthma, diabetes, psychiatric conditions, and cancer.

Model Organisms

Essential to the HGP were identical efforts to understand the genomes of various organisms commonly used in biomedical investigations, such as mice, fruit flies and roundworms. Such organisms were called "model organisms," because they serve as investigation models for how the human organism behaves.

The Genome

Children are taught in primary schools that everything is made up of atoms. Atoms combine to style molecules. For example two atoms of hydrogen and one atom of oxygen combine to form one molecule of water. How do atoms and molecules work unruffled to fashion the unique human being? All biological life is made up of an astonishingly intricate intermingled blend of molecules. They associate, break down and recombine into the same or a myriad of other forms of molecules. Every day trillions of actions and responses of molecules occur every second in progressions that provide energy, food and cell maintenance for our bodies. What type of instructions and communications tell these atoms and molecules what to do? The Human Genome Project aims to solve that mystery in a breath-taking detail that even the scientific world is awestruck.

The molecular structures in the nucleus of a cell (the primary unit of the body) called chromosomes are at the

fundamental level of the beginning of life. The chromosomes contain the genetic document- a chemical instruction set written in chemical code-that tell the human body how to arrange, structure, absorb and expel atoms and molecules. The totality of the genetic instructions is the **human genome**. Each individual has a inimitable genome, a specific chemical genetic instruction set. In reality, each individual is a genome.

Every individual receives one's genome at **conception.** A male sperm with its 23 chromosomes paired with a woman's 23 chromosomes in a fertilized egg, fashions a totally new human being which is beyond minds perceptive. Every individual starts life as a single cell. Beginning with that one cell a human being is made. Every growth pattern, stage and process of a human body occurs like clockwork-from foetal development to natal, infancy, childhood, adolescence and adulthood. For the first 18 years in life, on average one adds 100 million cells to the body *every minute.* Such impulsive growth is so perfectly programmed from your own set of instructions contained in that first cell that by age 20, one becomes an adult of more than 100 trillion (100,000,000,000,000) cells-segregated into heart, liver, spleen, bone, skin, muscle, stomach, intestines, eyes and most significant of all the brain. Scientists have estimated that 40 per cent of the genome is devoted to the development of the brain forlorn.

Design of Genetic Material

When scientists look into the design of the human cell they are wonderstruck to find it to be brilliant and its performance stunning. After 50 years the most marvellous biological furtive has been solved and that is how genes hustle all the development of the body's cells at the molecular level. To reach this considerate understanding they learned how the functions of the miniature cellular structure for feeding, repairing, eliminating waste, dividing and even vanishing and dying. With advances in technology, the magnificence of the structure of chromosomes was revealed.

The key to appreciating of the genes is the DNA, acronym for deoxyribonucleic acid. DNA is found in each cell's nucleus (hence it a nucleic acid), the command centre of the cell. DNA is also an instructional outline for every one of the 100 trillion cells that make up all body tissue. DNA directs each module of the cell in trillions of cellular processes that take place in the human body every second until death.

DNA structurally looks like ladder formation of two strands with stairs creating a double-helix shape. The ladder forms endure; massive molecule called the chromosome. Water has two atoms of hydrogen and one atom of oxygen and written as H2O, with a molecular weight of 18. The DNA chromosome molecule has a molecular weight of about 80 billion. DNA chromosome is a thin thread coiled in the cell's tiny nucleus and has a diameter of 2 nanometres. When stretched to full length it would be about one and a quarter inch long. The two DNA's strands appear like stilts made of a sporadic phosphate and sugar. The step like rungs between the strands are made up of paired bases of nitrogen compounds identified by the letters G, C, A and T (the first letters of the four kinds of bases: guanine, cytosine, adenine and thymine). These are the only substances in DNA; hence the genome consists of only these four-but in seemingly endless sequences. These compounds direct every cell in the human body what to do. Extraordinary bands of these compounds are our genes.

Genes

Fig 1: Human Genomes Researches through mapping

Genes are paired on each chromosome with sequences that account for specific traits and physical and physical characteristics. Each body trait requires one or more pairs of genes. For example the colour of eyes, shape of body parts and susceptibility to diseases all are found within the gene pairs of the genome. There are between 80,000 to 100,000 genes in the human genome. Scientists do not know where all the genes are, or even how many genes there are. The goal of the Human Genome Project is to decode everyone.

The universe is estimated to contain 100 billion galaxies with an estimated 200 billion stars each, and now scientists are realizing that each human body appears as complex and amazingly designed as the universe itself. For example the human brain has 100 billion neurons, with ineffable trillions of connections and patterns of boundless wiring sequences. We are unacquainted of what goes on in our cells as our genome tells our cells to assemble amino acids into proteins to make cell walls, and cell walls to split and divide and human beings are unaware of the constant stream of virtual miracles that keep human beings alive, alert and functioning.

Solving the puzzle

How did such an astoundingly complex process begin? How did the billions of atoms in each DNA molecule arrange themselves flawlessly for the self-endurance or what we call life? How did cells, DNA and chromosomes come about? Some claim that the greatest scientific proof that human beings were designed by a higher Power is this: The process of one genome creating a living, self-enduring organism cannot happen over time. It has to be right the first time, and it must entail literally billions of deliberate elements that must be in place and functioning perfectly, or else the cell

cannot exist and duplicate or for that matter replicate. The self-replicating cell exists only because its inherent scholarly systems- each involving billions of functions- interact perfectly. Otherwise it is deceased. The chromosomes and cells are extremely complex and beyond imagination that some scholars argue that they could never have evolved through random processes from nothing, even if given the endless time spans evolutionists require for their theory. Evolutionists are impotent to explain, for example, how and why heart tissue, liver tissue, skin and blood are distinctly different and have dramatically different functions. However, astoundingly, each cell contains the same DNA. Therefore a liver cell's DNA is alike to a brain cell's DNA. Still the mystery is how each cell knows its identity, function and locus in the body.

How do researchers create a genetic map?

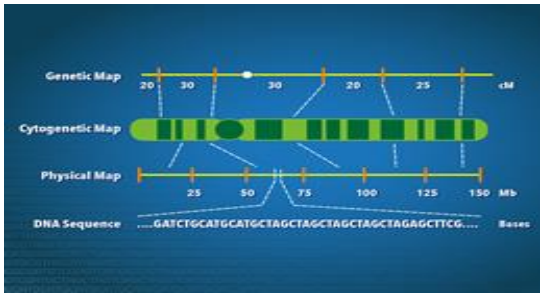

Fig 2: Example of a Genetic map

To produce a genetic map, researchers collect blood or tissue samples from members of families in which a convinced disease or trait is prevalent. Using various laboratory techniques, the scientists isolate DNA from these samples and examine it for unique patterns that are seen only in family associates who have the disease or trait. These characteristic patterns in the chemical bases that make up DNA are referred to as markers. DNA markers don't, by themselves, identify the gene responsible for the disease or trait; but they can tell researchers coarsely where the gene is on the chromosome. This is why: when eggs or sperm develop, the paired chromosomes that make up a person's genome exchange stretches of DNA. Think of it as a scuffling process, called recombination. The single chromosome in a reproductive cell contains some springs of DNA inherited from the person's mother and some from his or her father. If a particular gene is close to a DNA marker, the gene and marker will likely stay together during the recombination process, and they will likely be passed on together from parent to child. If each family member with a specific or certain disease or trait also inherits a particular DNA marker, it is very likely that the gene responsible for the disease lies near that marker. The more DNA markers there are on a genetic map, the more likely it is that at least one marker will be located close to a disease gene-and the easier it will be for researchers to zero in on that gene. One of the first major achievements of the HGP was to develop dense maps of markers spaced evenly across the entire human genome.

What are genetic markers?

Markers themselves customarily consist of DNA that does not contain a gene. Since markers can help a researcher locate a disease-causing gene, they are exceptionally valuable for tracking inheritance of traits over generations of a family. The development of easy-to-use genetic maps, coupled with the HGP's efficacious sequencing of the entire human genome, has greatly advanced genetics research. The improved quality of genetic data has reduced the time required to identify a gene from a period of years to, in many cases, a matter of months or even weeks. Genetic mapping data generated by the HGP's laboratories is freely accessible to scientists through databases maintained by the National Institutes of Health and the National Library of Medicine's National Center for Biotechnology Information (NCBI) [ncbi.nlm.nih.gov], as well as the Genome Browser of University of California, Santa Cruz.

Benefits of the Human Genome Project

Through decoding the human genome scientist's hope, among other things, to determine the grounds for many diseases, develop new treatments and cures and slow or reverse the aging process. Already researchers on the Human Genome Project have identified genetic disorders responsible for cystic fibrosis and some types of cervical, stomach and testicular cancer, among other diseases. A startling number of slight changes that make one person different from another--known as single nucleotide polymorphisms or SNPs -- have been identified. Until September 2000 more than 800,000 SNPs have been identified. Of the 800,000 now found, 300,000 were identified by the private-public SNP Consortium, which has accelerated its program The Consortium's members include Britain's Wellcome Trust, AstraZeneca PLC, Aventis Pharma, Bayer AG, Bristol-Myers Squibb Co., Hoffman-La Roche, Glaxo Wellcome Plc, Novartis Pharmaceuticals, Pfizer Inc., Searle (now part of Pharmacia), and SmithKline Beecham Plac and Motorola Inc., IBM, and Amersham Pharmacia Biotech. Other members include the Whitehead Institute for Biomedical Research at the Washington University School of Medicine in St. Louis, Missouri; the Wellcome Trust's Sanger Center, Stanford University's Human Genome Center, and the Cold Spring Harbor Laboratory in New York, New York. The **National Human Genome Research Institute (NHGRI**) has produced a series of fact sheets to provide an overview of the institute, explore the social implications of genetic research, and explain complex genetic concepts and research techniques to a non-scientific audience. Teachers, students and the general public alike will find the materials clearly written and easy to understand. The Human Genome Project also has its own moral and ethical issues. Parents will be motivated to abort unborn foetuses with anomalous genetic profiles. There will be an increasing trend towards fashioning "designer babies" by customizing personal genetic traits such as appearances, abilities, height, intelligence and eye and hair colour. Genetic acumen is a real leeway. Companies screen potential employees and deny jobs or insurance to those with genetic predisposition to some diseases. Finally, children could be sorted into communal classes or career tracks based on career potential.

"We have caught a glimpse of an instruction book (of life) Previously known to God." --Dr. Frances Collins, Director of the National Human Genome Research Institute, National Institutes of Health, Bethesda, Maryland.

Computational Analysis

Computers can be used very meritoriously to indicate the location of genes and of regions that control the expression of genes and to discover relationships amid each new sequence and other known sequences from many different organisms. This process is referred to as "sequence annotation." Annotation (the elucidation and description of biologically relevant features in the sequence) is the crucial prerequisite before the genome sequence data can become expedient, and the quality with which annotation is done will unswervingly affect the value of the sequence. In addition to substantial organizational issues, significant computational challenges must be addressed if DNA sequences that are produced can be successfully annotated. It is clear that new computational methods and a workable process must be implemented for effective and timely analysis and management of these data.

In considering computing related to the large-scale sequence analysis and annotation process, it is useful to examine previously developed models. Procedures for high-throughput analysis have been most notably applied to several microorganisms (e.g., *Haemophilus influenzae* and *Mycoplasma genitalium*) using relatively simple methods designed to facilitate basically a single pass through the data (a pipeline that produces a one-time result or report). However, this is too simple a model for analyzing genomes as complex as the human genome. For one thing, the analysis of genomic sequence regions needs to be updated continually through the course of the Genome Project—the analysis is never really done. On any given day, new information relevant to a sequenced gene may show up in any one of many databases, and new links to this information need to be discovered and presented. Additionally, our capabilities for analyzing the sequence will change with time. The analysis of DNA sequences by computer is a relatively immature science, and we in the informatics community will be able to recognize many features (like gene regulatory regions) better in a year than we can now. There will be a significant advantage in reanalyzing sequences and updating our knowledge of them continually as new sequences appear from many organisms, methods improve, and databases with relevant information grow. In this model, sequence annotation is a living thing that will develop richness and improve in quality over the years. The "single pass-through pipeline" is simply not the appropriate model for human genome analysis, because the rate at which new and relevant information appears is staggering.

CONCLUSION: We are now in a situation that we understand the benefits of human genome project, and so talking in the Indian context we have a large amount of scope, so that we unravel the new heights in genome discovery and genome mapping, utilising the Information technology advancements, and digital India concepts. This field has a variety of scope for teachers, researchers, doctors and computer scientists for further discovery into intricacies of human body and its truths of life. **Armed with the wealth of genetic data** generated by the Human Genome Project (HGP) and the state-of-the art resources available at the National Institutes of Health (NIH), clinical researchers at the National Human Genome Research Institute (NHGRI) are leading a new era in medicine - one where a more profound understanding of the biological basis of disease will pave the way for more effective ways to diagnose, treat and prevent illness. NHGRI's clinical research endeavors extend far beyond searches for disease genes. Among the many projects underway in intramural clinical research labs are:

a. Studies of a knock-in mouse having hereditary inclusion body myopathy.

b. Development of gene therapy approaches for X-linked severe combined immunodeficiency.

c. Conduct of clinical trials of a drug to combat mitochondrial disease.

d. Treatment of ACDC with bisphosphonates.

e. Determination of the safety and efficacy of a drug called nitisinone in the treatment of one type of albinism.

f. Pursuit of therapies to prevent progression of the muscle wasting and weakness in hereditary inclusion body myopathy.

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