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Human Genome Project: Expectations and Current Status

Abstract

Human Genome Project (HGP) was an international collaboration among various world scientists to come up with the sequence of complete human genome. There were many expectations associated with it as it was one of the largest projects in life sciences. Much hype was created from the beginning of this project like the discovery of various genes associated with severe diseases and understanding of their complexities would be very easier after the completion of HGP. It was thought that Genome sequencing will have a great impact on lives of people as it will help in better understanding of human evolution, development of novel therapies and disease diagnosis to discovery of novel drugs. This was also expected that in coming years, treatment for diseases like Alzheimer's, Parkinson's, diabetes and cancer would be possible by attacking their genetic roots. Although after a decade since the completion of HGP, researchers have tried to get many unsolved queries of human life but still many problems need to be solved in near future. In this review, we have summarized the major expectations and current status of HGP in recent scenario.

Keywords: Human genome project, Drug discovery, Genomics, Bioinformatics, Evolution

Mini Review

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Introduction

Around 64 years ago, a group of visionary scientists took biology into the molecular world [1]. The discovery of the structure of DNA helix was a major breakthrough in the field of biology which changed our basic perception and understanding of the flow of genetic information within the cell. This was the time when modern biology was born and a new branch of biology i.e. Molecular Biology was introduced. In the mid to late 1970s DNA cloning and Sanger's DNA sequencing methods were introduced which made scientists to dream that sequencing the whole human genome is possible. Many scientists believed in this concept of sequencing whole human genome, later called "Human Genome Project". They argued that the complete sequence of human genome would lead to a new understanding of mechanism of biological life and will benefit human health as well. Later in 1990s, the Human Genome Project started and took 13 years to sequence the entire DNA content of human cells [2].

Expectations and Outcomes of HGP

The Human genome project benefits the human life in many aspects. It helps to understand the evolution of human being, to identify the diseases causing genotypic change in human which helps to direct treatment in an appropriate way, to identify oncogenes and related mutations which lead to different forms of cancers. The whole genome sequence is publically available at nucleotide sequence repository of National Center for Biotechnology Information (NCBI). Two other fields of biology evolved after the genome sequences were made available, these are Comparative Genomics and Bioinformatics. The principles of Comparative genomics are very straightforward [3]. It aims to find the conserved regions in the genome during the time of evolution and helps to construct the phylogenetic tree based on the inter species genomic distances. It is not only aims to discriminate conserved regions of DNA from divergent and functional from

nonfunctional DNA [4,5], but also helps in identifying the general functional class of certain DNA segments, such as coding exons, noncoding RNAs, and some gene regulatory regions. At the other hand, Bioinformatics aims to the systematic development and application of IT solutions to handle biological information by addressing biological data collection and warehousing, data mining, database searches, analysis and interpretation. Several tools have been developed for biological data analysis for example BLAST to find out similar nucleotide/amino-acid sequences, ClustalW to align two or more nucleotide/amino-acid sequences, Primer3 to design primers probes for PCR techniques, etc. Hence the technology and resources generated by the Human Genome Project and other genomics researches have already produced a major impact on life science research. Currently worldwide Biotechnology research poses large opportunities; and more than \$100 billion have been invested in several commercial and scientific researches in the biotechnology industry. Some of the current and genome based research include:

- I. Development of molecular medicine, which is possible only because of the availability of detailed genome maps that helped researchers to identify the association of genes with dozens of genetic disease, including Alzheimer's disease, Fragile X syndrome, Myotonic dystrophy, inherited colon cancer, Breast cancer, etc.
- II. Understanding of human genome gave an enormous impact on our ability to determine the risks posses by individuals to any toxic agents. A slight genetic difference makes few people more susceptible and others more resistant to such agents. Many research works are going worldwide to determine the genetic basis of such variability. These researches also include the understanding of effects of low-level exposures to radiation and other energy-related agents, especially in terms of cancer risk [6].

- III. One more major focus of genomic research is to understand human evolution and the common linkage with all other form of life. Comparative genomics studies between humans and other organisms have already led to determine similar genes association and helped to calculate the inter species distances in order to construct the phylogenetic tree. Further comparative studies will help to determine yet-unknown function of thousands of other genes.
- IV. The area of DNA Forensics has emerged as a surprising way to identify the crime suspect with small amount of DNA sample from crime scene. Not only human but any organism can be identified by examining the DNA sequences. As we know that our genome is 99.99% same but still the variation among individual is because of those 0.01% unique dissimilar regions of genome. To identify individuals, forensic scientists scan about 10 DNA regions that vary from person to person and use the data to create a DNA profile of that individual. There is extremely rare chance that two persons have the same DNA profile for a particular set of regions [7].

Except these, there are large numbers of other major genomic researches are going worldwide to achieve different goal. For example a very challenging research is going on to create a new Energy sources using microorganism called biofuels and to monitor environment to detect pollutants and to find a safe, efficient environmental remediation technique using microbes [8]. This is the knowledge of complete genomic sequence that today we know that all the proteins and several RNA which acts to perform various cellular functions are encoded in these genomic sequences [9]. All these research areas are directly linked to human life and aims to benefit human society. Today all these research works are only possible because of the successful completion of Human genome project and availability of whole genome sequence and the complete genome map. So we can say that, this project has lived up to the expectations of scientists and day by day still creating new research opportunity in the field of life-science.

Role of HGP in Drug Design/Development

A group of scientists believed that a potential breakthrough in the field of drug design was only possible after the successful completion of Human Genome Project. The HGP helped scientists to understand the Physiology of genetic diseases including cancer, heart disease, diabetes, hypertension, etc. The availability of whole genome sequences and various advance computing tools to visualize, analyze the DNA sequencing database and to predict three-dimensional protein structure and function actually made a breakthrough and made structure-based drug design possible. HGP also helped in identification of potential genetic changes, called mutation which creates the risk of any genetic disorder, and to develop strategies for specific drug targeting to eliminate these mutations [10].

The identification of new, clinically relevant, molecular targets is of most importance to the discovery of innovative drugs. It has been estimated that up to 10 genes contribute to multifactorial diseases. Typically these disease causing genes are linked to other 5 to 10 gene products in physiological circuits which are also

suitable for pharmaceutical intervention. If these numbers are multiplied with the number of diseases that pose a major medical problem in the industrial world, then there are ~5000 to 10000 potential drug targets. With the help of Bioinformatics techniques one can also compare the entire genome of pathogenic and nonpathogenic strains of a microbe and identify genes/proteins associated with pathogenesis. So the genomics and bioinformatics studies of genomic sequences can help in identifying large number of potential disease targets.

After target identification the next step is to find the lead compound which can bind to the target and regulate their activity. In the area of structure based drug designing, three dimensional structures of compounds from virtual or physically existing libraries are docked into binding sites of target proteins with known or predicted structure. The scoring functions evaluate the steric and electrostatic complementarity between compounds and the target. The highest ranked compounds are then suggested for biological testing. This method effectively reduces the overall drug discovery time period.

The availability of the genomic sequence also helps in determining the toxicological effect of certain compound on any individual. Small genetic difference makes few people more susceptible and others more resistant to certain agents. Hence a drug may act differently on to different individual. So by analyzing the genomic makeup of individual we can predict these aspects.

These days structure based drug designing is a main choice of any drug discovery project [11]. Several drugs are already available in the market using this technique, for example: Anti HIV drugs, Anti cancer drugs like Imatinib, Drugs for neurological diseases like Rituximab, several antibiotics, etc. So HGP leads the area of pharmaceutical science to a new direction of more effective and more cost effective drug designing with the help of structure based drug designing approach.

Conclusion

HGP was for many reason one of the largest collaborative projects of entire world at that time. Many scientists believed that it would revolutionize the biological/medical field. Over the time it gave rise to many novel dimensions to understand the complexities of human life. May different branches of life-science emerged after the completion of this project aiming to decipher the hidden information from human genome. Genomics and Bioinformatics are the two areas of which the maximum focus is on understanding the human genome and to develop computational tools in order to make the work easier. HGP also helped a lot in the understanding of the mechanism of various diseases. It helped scientists to look for various new targets and active lead compound in order to cure the diseases. Very vast works are going worldwide on various diseases to find the better cure and all of these are happening because of the presence of entire human genome sequence with us. Hence we can conclude that HGP defiantly lived up to the expectations of scientific community.

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