Introduction

Scientists over the time have explored and mapped lands, oceans and space with the anticipation of escalating our knowledge of the environment in which we live and sustain. Underlying this exploration for knowledge is also the keen and acute craving to advance and progress the human subsistence and existence through the discovery of beneficial material resources.1 The Human Genome Project (HGP) is an international scientific project to decode sequence and map the human genome, the blueprint of life, as well as to document humanities genetic resources and information.2

The HGP involves the discovery and sequence of the full DNA complement in a single human somatic cell. Its primary goal is a listing and location of our genes -- the single unit of heredity responsible for how we develop from conception, how we grow and mature, how we live, and how we die.3 The HGP, which can be compared with the astounding achievements like landing on moon and space by humans, has the prospects to transform the medical and health aspects. No wonder some people has described HGP as the “Holy Grail of Biology” and a mission to

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2 <http://www.ornl.gov/sci/techresources/Human_Genome/project/about.html> the site is maintained by U.S. Department of Energy Office of Science, Office of Biological and Environmental Research, Human Genome Program(Last visited on 12-1-2008).
3 Supra note 1, See Marion at 46.
understand all about our existence which reveals the “Book of Man”. At the same time it has the potential to create perpetual, permanent, and structural inequalities in the society. In order to properly understand the nature and breadth of these problems and issues, a comprehensive understanding of the HGP is required. The chapter will analyze the origin, development, importance and present status of the project. A brief analysis of the potential benefits and ramification of HGP will also be undertaken.

Origin and Development of the HGP

The human genome Project: the crown jewel of 20th century biology, heralded at the White House, plastered on the covers of countless magazines—and at last spelled out in intricate detail in both Science and Nature⁴ is an international collaboration of the world’s best scientific minds created to identify the form and content of human genome.⁵

An analyzes of the beginning of Human Genome Project brings forth that it did not have a sudden beginning, rather it was a natural culmination of the growth in biology the world witnessed specially after the discovery of genes by Watson and Crick.⁶ Following the discovery of genes as a hereditary unit, in 1954, George Gamow identified that DNA sequence was a four-letter code embedded in the order of base pairs.⁷ Later in 1975, Fredrick Sanger had successfully demonstrated that double

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stranded DNA could be sequenced using deoxy sequencing technique.\(^8\)

Alan Maxam of Harvard also independently developed a completely different method to determine the order of those base pairs that same year. This method was pronounced to the scientific world at various scientific conferences and seminars. The earliest and first practical prototype of this method developed under the direction of Lloyd Smith, at the California Institute Technology in 1986. It was later applied commercially by Applied Biosystems, Inc., in 1987.\(^9\) These seminal technologies gradually led to the genome project.

By 1978, it was becoming apparent that these genetic sequence information needed to be catalogued systematically to make it useful to the scientific community. After several years of intense, often tense and acrimonious discussion, twin databases were established under the European Molecular Biology Laboratory in Heidelberg and as GenBank at Los Alamos National Laboratory.\(^10\) The explosion of minicomputers in the 1970's and microcomputers in the 1980's fuelled the attention to DNA sequence information because computational methods were obviously the only way to analyze the deluge of DNA sequence information produced by sequencing techniques. The technologies were thus present, but it took the spark of an idea of using them as part of a large organized effort to ignite the fire, out of which rose the Human Genome Project.\(^11\)

One of the first to grasp that potential of decoding and sequencing of human genome was Robert Sinsheimer, a biologist the then Chancellor of the University of California (UC), Santa Cruz. At that time The Hoffman

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\(^10\) Ibid. Walter Goad of the Theoretical Biology and Biophysics Group at Los Alamos National Laboratory and others established the Los Alamos Sequence Database in 1979, which culminated in 1982 with the creation of the public Gen Bank funded by the National Institutes of Health.

\(^11\) Supra note 7, See Robert, at 169.
foundation offered a $36 million to the University of California for the construction of a new telescope for its world renowned astronomy department. Sinsheimer was looking for a project of similar magnitude in biology. At the time, the largest genome yet sequenced was the minuscule Epstein-Barr virus—and that feat had taken several researchers years to complete. To apply such tools to the human genome, nearly 20,000 times bigger at 3 billion bases, was audacious beyond belief. In 1985, Sinsheimer assembled some of the best minds in the nascent field of genome analysis to hash over the proposal, but their idea for a genome sequencing Institute though seemed bold, captivating and exciting, it was not feasible mainly due to the funding and lack of technology. The idea died in its natural course but before that it had captured Gilbert's imagination. Gilbert soon became the proposal's biggest champion. Gilbert infamously called it the quest for biologist" holy grail" and he soon won over another giant of molecular biology: James Watson, who shared a Nobel Prize with Francis Crick and Maurice Wilkins for their 1953 discovery of the double helical structure of DNA.

Without knowing about the Santa Cruz workshop, Renato Dulbecco of the Salk Institute conceived of sequencing the genome as a tool to understand the genetic origins of cancer. Dulbecco, a Nobel Prize winning molecular biologist, laid out his ideas on Columbus Day, 1985, and subsequently in other public lectures and in a commentary for Science. He advocated that a large-scale program rather than a piecemeal

13 Id., at 18.
14 Gilbert along with Allan Maxam, of Harvard University, had invented a brand-new technique that enabled scientists for the first time to determine the genetic sequence of an organism. For this Gilbert went on to share the Nobel Prize with Fred Sanger of Cambridge University, who independently invented a similar technique.)
15 Supra note 12. See Witkowski at 13.
approach was the best way to make progress in the war against cancer, launched by President Nixon in 1971. A project to sequence a complete genome would be important for the study of all disease and development, not just cancer and the obvious place to start was with the human genome.\textsuperscript{17} His concluding remarks still resonate

"Its significance would be comparable to that of the effort that led to the conquest of space, and it should be carried out with the same spirit, Even more appealing to make it an international undertaking, because the sequence of one might be tempted to believe that this genetic blueprint might be the closest mankind has come to grasping the Holy Grail. If current trends are anything to go by, the technologies of the not-so-distant future might well enable mankind to tinker with DNA to the extent of playing God. Humanity has to face the situation of being responsible for tasks, which will increasingly put his own survival as a species at risk. More importantly, there are the lingering questions on the privacy of individual genetic data, fairness in evaluation of genetic facts, consent before use of such data, exploitation of rare genes in individuals, patenting of genetic material, conduct in reporting results in population genetics, genetic testing of modified DNA on individuals among others. The human DNA is the reality of our species, and everything that happens in the world depends on those sequences."\textsuperscript{18}

That conviction stemmed from having seen, first, the tremendous advantages of knowing the sequence of SV40 in 1978 and adenovirus genomic DNAs in 1979–1980, particularly for deciphering their biological properties. In each of these instances, as well as for the longer and more complex genomic DNAs of the herpes virus and cytomegalovirus, knowing the sequences was critical for accurately mapping their mRNAs, identifying the introns, and making pretty good guesses about the

\textsuperscript{17} Kevin Davies, \textit{The Sequence, Inside the Race for the Human Genome}, at 11, (2002).
\textsuperscript{18} \textit{Ibid.}
transcriptional regulatory elements. Even more significant was the ability to engineer precisely targeted modifications to their genomes. One could easily imagine that knowing the human DNA sequence would enable us to manipulate the sequences of specific genes for a variety of hitherto undoable experiments.19

Dulbecco's commentary in science catapulted the concept of a Human Genome Project into the scientific mainstream; the ambitious idea had also captivated Charles-DeLisi, a cancer biologist who was then head of the Office of Health and Environmental Research at the Department of Energy (DOE).20 To DeLisi, the genome project was a logical outgrowth of DOE's mandate to study the effects of radiation on human health.21 The DOE Office of Biological and Environmental Research (OBER) of DOE and its predecessor agencies the Atomic Energy Commission and the Energy Research and Development Administration have long sponsored research into genetics, both in microbial systems and in mammals, including basic studies on genome structure, replication, damage, and repair and the consequences of genetic mutations.22 The DOE had been studying survivors of the atomic bomb explosions in Japan Perhaps Another equally or more compelling rationale, but one which was never openly talked about was that a large and massive scale new scientific endeavour could provide new focal for research laboratories, whose nuclear atomic and bomb making skills were in diminishing demand.23

At the urging of DeLisi, the Los Alamos National Laboratory hosted a workshop in Santa Fe, New Mexico, in March 1986 just a few days before the publication of the Dulbecco's commentary. The idea of sequencing Human genome was hotly debated and scientific excitement was clearly

20 Id., at 1184.
21 Id., at 1183.
22 Supra note 12, See Witkowski at 17.
23 Id., at 23.
visible.24 But most participants concluded that sequencing should be done only after a physical map of the genome had been assembled. David Botstein, The Stanford University geneticist who first proposed a genetic map of human DNA in 1980 chastised the initiative as DOE's program for unemployed bomb makers.25 The idea quickly gained momentum. Two months after Dulbecco's controversial call for a big science genome project legions of DNA dignitaries gathered at the Cold Spring Harbour Laboratory, in New York for a meeting entitled "The Molecular Biology of Homo Sapiens" dominated the proceeding in the next meeting at a June meeting at Watson's Cold Spring Harbour Laboratory in New York.26 By then, biologists are beginning to think the project just might be possible. But another question equally important was whether it was worth doing was another matter.27

The scientific value seemed dubious as well. Although many biologists agreed that maps of the chromosomes would be useful for finding genes, what good would come from deciphering every A, T, G, and C, especially since most of them were "junk" that did not code for genes. The sequence might be handy to have, but "was it worth the cost, not in terms of dollars but in terms of its impact on the rest of biological science.28

Political posturing continued until 1988, a special national research Council panel chaired by Bruce Albert's, in USA deliberated on the wisdom of the Human Genome Project.29 Its final report advocated the international program led by the United States to sequence the human genome. With the cost of DNA sequencing obscenely high, the panel

24 Supra note 17, See Kevin, at 13.
25 Id., at 15.
26 Ibid.
28 Supra note 4 See Roberts, at 1183.
recommended the postponement of DNA sequencing until improved technology had driven down the cost probably in five years. Instead it was decided that early efforts should focus on mapping the human genome and characterizing the genomes of other organisms, such as the mouse, the fruitfly, etc. Thus finally the National Research Council (NRC) panel gave the project its official seal of approval calling for a rapid scale-up in “new and distinctive” funds to $200 million a year over the next 15 years.

In the process, the panel redefined the project, laying out a phased approach that mollified critics and has guided the initiative ever since. Rather than plunge into sequencing—which no one knew how to do on a massive scale anyway—the project should begin by constructing maps of the human chromosomes. These would greatly speed the search for disease genes, offering immediate medical payoffs. Soon thereafter, the National Academy of Sciences convened a blue-ribbon committee, many members of which had been among the critical voices at CSH. Their report recast the scope and direction of the project in a more constructive way; the principal change was the proposal to proceed in phases: determine the genetic map by use of principally polymorphic markers, create a physical map consisting of linked cloned cosmids, and focus on developing more cost- and time-efficient means of sequencing DNA. The most important recommendation was to include in the project the sequencing of the then favourite model organisms: Escherichia coli, Saccharomyces cerevisiae, Drosophila melanogaster, Caenorhabditis elegans, and the mouse. It also provided a livelihood for those interested in mapping their favourite organism and for those committed to cloning

31 Supra note 17, See Kevin at 17.  
32 Supra note 4, See Roberts at 1183.  
and mapping large segments of DNA. In the end, people were mollified by the realization that they would not be left out of the project’s funding. Also, the proposal had logic for how to proceed and the acceptance that useful information would be generated long before the project was completed.34

Meanwhile, one of the most ardent supporters of Sinsheimer and Delbecco’s proposal, and a member of NRC, Gilbert proposed a blind sequencing project randomly cutting the genome, sequencing the DNA fragments and reassembling them.35 He announced that he would form a private corporation that would sequence the genome, obtain a copyright on the data, and then sell it to researchers.36 But his vision of sequencing the entire genome drew an openly hostile response. The main criticism was the huge cost which will necessarily deny fund for many worthy projects. Indeed the late 1980s the proportion of the grants funded by the National Institute of Health (NIH) fell from 40 percent to less than 25 percent.37 Critics maintained, the cost of doing this project would diminish federal funding for individual investigator-initiated science and thereby would shift the culture of basic biological research from “Little Science” to “Big Science.” Some feared that biology would experience the same consequence that physics did when massive projects like the Stanford Linear Accelerator Centre were undertaken in that field. Many thought that Gilbert’s approach was boring and thus would not attract well-experienced people, which, most likely, would make the product suspect. Moreover, the benefits of the sequence project might not materialize until the very late stages. Gilbert however, was impatient with the panel’s cautious approach and with the interagency dithering. Arguing that the technology was already good enough to sequence the human genome, he left the NRC panel to launch his own company, Genome Corp. His plan, remarkably similar to J. Craig Venter’s vision a decade later, was to set up a

34 Ibid.
36 Id. at 12.
37 Supra note 17, See Kevin at 16.
sequencing factory to churn out the data, which he intended to copyright and sell. "[It will be] available to everyone ... for a price," he explained.\textsuperscript{38} The plan infuriated Watson, who rankled at the idea of selling something as fundamental as data on human DNA. But the debate subsided when Gilbert failed to raise sufficient funds. But Gilbert had much more success when a few years later, when he co founded Myriad shotetics, the Salt Lake City company 1994.\textsuperscript{39}

The blue ribbon committee of National Academy of science finally recommended the government funding of $ 200 million per year up to fifteen years. The only thing it refused to voice an opinion was whether the genome project should be administered by the Department of Energy or the NIH. By this time DOE was pushing ahead with its genome initiative. The plan survived a congressional hearing in March 1987. and DeLisi submitted his first budget, for $12 million, in 1988.\textsuperscript{40} As the genome project gained congressional funding and scientific respectability, NIH wrested control from DOE. Urged on by a group of advisers who met outside Washington, D.C., in Reston, Virginia, in March 1988, then-NIH director James Wyngaarden announced that NIH would create a special office for genome research.\textsuperscript{41} In short order, he nabbed Watson to head it, and with that, NIH was firmly ensconced as the lead agency. Watson along with Wynagaarden appeared for the press conference to announce that the Human Genome Project would begin officially in 1990. surprisingly this was not the only thing he announced at the press conference. The one aspect that is directly related to the topic of this meeting is that he stated that some 3 percent to 5 percent of the budget for the centre would be devoted to the studies of ethical and legal issues arising out of Human genome Project.\textsuperscript{42} It was a sincere effort to ensure that society was prepared for

\textsuperscript{38} Leslie Roberts, "Watson may Head Genome Office", \textit{Science}, at 878, 13 May (1988).
\textsuperscript{39} Supra note 17, See Kevin at 16.
\textsuperscript{40} Id., at 17.
\textsuperscript{41} Supra note 35, See Roberts, at 1185.
\textsuperscript{42} Supra note 12, See, MGK Menon, at 19.
the tidal waves of information on the horizon. Recalling the Nazi atrocities, against gypsies and the mentally ill, Watson solemnly wrote

"We need no more vivid reminders that science in the wrong hands can do incalculable harm." Watson skilfully deflected scientific criticism of the project while searching for political support. "Once NIH got interested, many more people became involved. In October 1989, Watson's unit received its new status as the National Centre for Human Genome research and a budget for the fiscal year 1990 $60 million; soon after, an understanding was reached with DOE about how these two agencies should divide up the genome pie. Following a meeting at Banbary Centre at Cold Spring Harbour Laboratory, it was decided that the National Laboratories of the DOE should use their technological expertise to concentrate on developing novel strategies for dealing with genome size DNA. And while the centre for genome research would concentrate on more biological needs, such as new approaches to gene mapping. DOE and NIH signed a Memorandum of Understanding in October 1988 to coordinate the activities aimed at characterizing the human genome." Following a meeting at Banbary Centre at Cold Spring Harbour Laboratory, it was decided that the National Laboratories of the DOE should use their technological expertise to concentrate on developing novel strategies for dealing with genome size DNA. And while the centre for genome research would concentrate on more biological needs, such as new approaches to gene mapping. DOE and NIH signed a Memorandum of Understanding in October 1988 to coordinate the activities aimed at characterizing the human genome. Through great effort and expense, scientists in molecular biology, biochemistry, math, computer science, engineering and the health care industry have worked together to turn what began in 1985 as a simple campus improvement project at the University of California, Santa Cruz into an international scientific consortium. This cooperative effort now known as the Human Genome Project, begun in 1989, and officially kicked off in 1990.

44 Supra note 12, See MGK Menon at19.
46 Supra note 1, See Maria at 46.
47 Supra note 17, See Kevin at 21.
The genome project embraced three main technical goals: genetic linkage maps to trace the inheritance of chromosome regions through pedigrees; physical maps of large chromosome regions, to enable the direct study of DNA structure in search of genes; and substantial DNA and substantial DNA sequence information, enabling the correlation of DNA changes with alterations in biological function. If history were logical, then the genome project would have grown from a discussion of each in turn and how to bring them together into a coherent plan. History is not logical, however, and it was DNA sequencing technology rather than genetic linkage mapping that gave rise to the idea of a human genome project. 48

Although Watson’s peerless stature would inevitably spin off the genome project helped garner support for the program many scientists’ still harboured reservations. Leading the opposition was Bernard D. Davis and two dozen fellow faculty members who denounced the politically unstoppable initiative arguing that just as the known sequences of a few viruses had not had a profound effect on understanding viral biology, neither would the complete sequence of the human genome transform human biology.49 But Watson’s real troubles came in the form of new NIH Director Bernadine Healy, the two got entangled in a fight over the issue of gene patenting what sparked the fight was NIH move to seek patents for hundreds of gene fragments identified by NIH Scientist Craig Venter. Watson quit Genome Project abruptly on April 10, 1992.50

After Watson’s sudden departure, NIH picked gene hunter Francis Collins of the University of Michigan, Ann Arbour, to take the helm. A physician by training, Collins brought a different perspective to the genome project, placing its medical applications front and centre. 51

48 Supra note 7, See Robert at 1184.
49 Supra note 17, See Kevin at 30.
50 Id., at 31.
51 Supra note 35, See Roberts at 1183.
Venter, a biologist with the NIH, who had earlier sparked controversy over the issue of patenting too, left NIH in 1991 when he was offered $70 million from a venture capital company, The Institute for Genomic Research (TIGR) to try out his gene identification strategy at a new non-profit. Later he went on to form Celera Genomics Corporation. In 1998, Craig Venter, announced his participation in an independent genome-sequencing project planning to decode the entire Human Genome by the year 2001. The HGP and Venter worked independently, competing to be the first to describe the entire human genome sequence. In 2000, the British Prime Minister, Tony Blair, and President Clinton announced that all genomic information should be free, which led to the posting of any newly discovered sequences on the internet. In addition, Clinton and Blair expressed their desire for the two groups to work together on decoding the human genome. Subsequently, deliberations took place between Collins and Venter on this issue, and on June 26, 2000, both of them jointly released the rough draft of the human genome. Critics express several concerns about the Human Genome Project (HGP), and most involve the extent of the project or its funding. Original proposals for the project emphasized sequencing the entire human genome. This goal, however, is controversial because of the high cost and because many critics believe that sequencing a huge amount of non-coding DNA should have low priority in a time of limited funds for research. On the other hand, most individuals involved in the project agree that detailed genetic and physical maps would be extremely useful. Therefore, mapping of the genome now is the primary goal, with complete sequencing to follow only if the cost becomes reasonable.

Only about 5 percent of the genome contains sequences that are coding regions, and some biologists still maintain there is little point in sequencing the other 95 percent. South African Sydney Brenner, one of the founding fathers of the modern genetics era, had argued from the very beginning that DNA sequencing should focus first on the small percentage of the genome that contains genes, the far less interesting “junk DNA” later. He stressed that he was not against the project in principle. Because biologists already know that several regulatory signals are in noncoding regions of DNA, a compromise has been reached. A few pilot sequencing projects are focusing on sequencing certain coding regions that are most likely to contain information valuable to the medical and biological communities.

Object and Goals of Human Genome Project

A set of specific goals for the 15-year HGP had by 1990 been formulated and presented to the congress in USA. This can be divided into major two phases. The first phase covering year 1993-1998 and second when goals were revised from 1998-2003. In the initial phase Five-year goals have been identified jointly by NIH and DOE for the following areas, which together encompass the Human Genome Project: There were two major objectives. The first was to develop detailed maps of the location of genes in the human genome and the genomes of several other well-studied organisms such as Bacteria, Yeast, Nematode, Fruit fly (Drosophila), Mouse and

54 Supra note 17, See Kevin at 16.
55 Supra note 53.
It was anticipated that knowledge gained by the study of genomes of other organisms would assist in the analysis of the human genome. The second objective was to determine the sequence of the coded information contained in the DNA of the various genomes studied and from this to identify all of the estimated 30,000 human genes. This information is in the form of chemical “bases” that are described by the letters A, T, G and C. This plan sets out specific scientific goals to be achieved in the first five years together with the rationale for each goal. The specific goals will be reviewed annually and updated as further advances in the underlying technology occur.

Since most of the earlier goals were achieved, a new plan, for 1998-2003, became operational, in which human DNA sequencing became the prime focus besides this major goal other consisted of

- Data Collection and Distribution
- Ethical, Legal, and Social Considerations of Genome Research
- Research Training of Genome Scientists
- Technology Development of Sequencing
- Technology Transfer

Genome Sequencing

Providing a complete, high-quality sequence of human genomic DNA to the research community as a publicly available resource became the HGP’s highest priority goal after 1998. The enormous value of the human genome sequence to scientists and the considerable savings in research costs its widespread availability will allow was the compelling reason

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behind this goal.\textsuperscript{59} This was definitely a highly ambitious, even audacious goal, given that only about 6\% of the human genome sequence has been completed in the year of 1998.

Technology Development Sequencing

Since the commencement of Human Genome DNA sequencing technology and the amount of sequence produced every year had increased rapidly. Even then there was a dire requirement of more efficient sequencing technology for sequencing of additional genomes, comparative sequencing of closely related genomes, and sequencing to assess variation within genomes for biological and medical research.\textsuperscript{60} Thus increasing the throughput and reduce the cost of current sequencing technology through increased automation, miniaturization, and integration of the approaches currently in use, together with incremental, evolutionary improvements became another important goal of the second phase of HGP.\textsuperscript{61}

Technology for Functional Genomics

The HGP revolutionized the way biology and medicine will be explored in the next century and beyond. This became possible through the availability of entire genome sequences which has enabled a new approach to biology called functional genomics. Therefore, identifying and sequencing a set of full-length DNAs that represent all human genes and model organisms was given high priority in the second phase.\textsuperscript{62} Further goals included research on methods for studying functions of nonprotein-coding sequences and develop technology for comprehensive analysis of gene expression.

\textsuperscript{59}\url{www.nhgri.nih.gov/98plan/elsi//cgi/content/full/282/5389/682/F3} (last visited on 6—02-2008).
\textsuperscript{60} Ibid.
\textsuperscript{61} F. Collins and D. Galas, Science 262, 43, (1993).
\textsuperscript{62} Id., at 44
Comparative Genomics

Because all organisms are related through a common evolutionary tree, the study of one organism can provide valuable information about others. Comparisons between genomes that identify other model organisms that can make major contributions to the understanding of the human genome and support appropriate genomic studies. Hence sequencing of other organisms emerged as a major priority during the year 1998-2003.

Ethical, Legal, and Social Considerations of Genome Research

While recognizing that genetics is not the only factor affecting human well-being, the NIH and DOE were acutely aware that advances in the understanding of human genetics and genomics will have important implications for individuals and society like issues of privacy, discrimination, eugenics determinism etc Examination of these ethical, legal, and social implications (ELSI) of genome research therefore, became an integral and essential component of the HGP. This attention was clearly visible from the first press conference of Watson where he announced that 3 to 5 percent of the genome research will be earmarked for studying ethical and legal issues arising out of the project. In a unique association, biological and social scientists, health care professionals, historians, legal luminaries, academicians and others became committed to the evaluation of these issues. The major ELSI goals were

a) Examine the issues surrounding the completion of the human DNA sequence and the study of human genetic variation.

b) Examine issues raised by the integration of genetic technologies and information into health care and public health activities.

c) Examine issues raised by the integration of knowledge about genomics and gene-environment interactions into no clinical settings.

64 Ibid.
d) Explore ways in which new genetic knowledge may interact with a variety of philosophical, theological, and ethical perspectives.
e) Explore how socioeconomic factors and concepts of race and ethnicity influence the use, understanding, and interpretation of genetic information, the utilization of genetic services, and the development of policy.  

Training

The HGP created the need for new kinds of scientific specialists who can be creative at the interface of biology and other disciplines, such as computer science, engineering, mathematics, physics, chemistry, and the social sciences. Developing and training such scientists become one of the major goals of HGP

Mapping the Genome: Progress and Present Status of the Human Genome Project

The Human Genome Project was designed to be a worldwide effort, with about two-thirds of the work to be handled by University and Government groups in the United States, the remainder by United Kingdom, France, Germany, and Japan. Began formally in 1990 the HGP was planned to last for 15 years, Thousands of scientists, working in more than 100 laboratories and different countries around the world, have contributed to the Human Genome Project since its inception. Thanks to the development of later generations of high-speed automatic sequencers and supercomputers to handle the enormous amount of data generated, work on the project progressed well ahead of schedule and well under budget, accelerating the completion date to 2003. The major landmarks during this

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The Human Genome Project: Its Relevance, Triumphs and Challenges

process were on December 2, 1999, more than 100 scientists working together in laboratories in the United Kingdom, Japan, the United States, Canada, and Sweden announced the first completely sequenced human chromosome 22, the smallest of the autosomes. To assure the accuracy of the sequenced data, each segment was sequenced at least ten times.68

Next landmark was the publication of the human genomic map. On Monday, June 26 2000, the then US President Bill Clinton strode into the east room of white house, followed by two proud men. Craig Venter and Francis Collins. He announced “Today we are learning the language in which god created life We are gaining even more awe for the complexity, the beauty, the wonder of gods most divine and sacred code, Clinton added “Modern science has confirmed what we first learned from ancient faiths, the most important fact of life on this earth is our common humanity.69 It was followed by a Sequence tagged Sites (STS) based map and then by a transcript map. A breakpoint map highlighting the chromosomal rearrangements that recur frequently and an STS-based radiation hybrid map appeared soon thereafter.70

The International Human Genome Sequencing Consortium published the first draft of the human genome in the journal *Nature* in February 2001.71

With the sequence of the entire genome’s three billion base pairs some 90 percent complete. The Human Genome Project international consortium announced that 2 billion of the 3 billion “letters” that constitute the genetic instruction book of humans have been deciphered and deposited into Gen Bank.72

The public database of DNA sequence operated by the National

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69 Supra note 17 See Kevin at 236.
72 “Two Thirds of Human DNA Script Deciphered by Human Genome Project; Public Consortium To Complete "Working Draft" In June, Science Daily, April (2000).
Institutes of Health is accessible freely and without restrictions to all scientists in industry and academia.\textsuperscript{73}

The Draft of HGP has revealed that there are probably somewhere between 30,000 and 40,000 human genes. The human genome was estimated to contain 3.15 billion bases. The consortium sequenced more than 22 billion pairs of DNA. Celera calculated that human genome contains 3.12 billion bases, some 30 million bases fewer than the public consortiums estimate. Celera sequenced 26.4 million DNA segments of about 550 letters a piece. The total sequencing was 14.5 billion letters providing 4.6 times coverage of the genome.\textsuperscript{74} The high-quality reference sequence was completed in April 2003, marking the end of the Human Genome Project two years ahead of the original schedule. Coincidentally, this was also the 50th anniversary of Watson and Crick's publication of DNA structure that launched the era of molecular biology.\textsuperscript{75}

Sequencing the human genome has led to some surprising results. For example, we once thought that highly evolved humans would need a great many genes to account for their complexity, and scientists originally estimated the number of human genes to be about 100,000. The draft of the human genome, however, indicates that humans may have only about 30,000 genes, far fewer than originally expected. Indeed, this is only about one-third more than the number of genes found in the lowly roundworm, \textit{Caenorhabditis elegans} (approximately 20,000 genes), and roughly twice the number of genes in the fruit fly \textit{Drosophila melanogaster} (approximately 14,000 genes). Subsequent estimates have placed the number of human genes closer to 70,000; the true number is unknown as of mid-2007. Scientists have learned that most of the genome does not

\textsuperscript{73} Available at http://www.ncbi.nlm.nih.gov/genome/seq) (Visited on 3-08-20008
\textsuperscript{74} Supra note 17, at 236.
code for proteins, but rather contains "junk DNA" of no known function. In fact, only a small percentage of human DNA actually encodes a gene.\textsuperscript{76}

The full sequence was completed and published in April 2003.\textsuperscript{77} The completed human sequence can now identify their locations. This ultimate product of the HGP has given the world a resource of detailed information about the structure, organization and function of the complete set of human genes. This information can be thought of as the basic set of inheritable "instructions" for the development and function of a human being.\textsuperscript{78} Available to researchers worldwide, the human genome reference sequence provides a magnificent and unprecedented biological resource that will serve throughout the century as a basis for research and discovery and, ultimately, myriad practical application.

The project evolved over the 90s decade and in its final stages had over 16 international centres involved globally

- Baylor College of Medicine, Houston, Texas, USA
- Genoscope, Evry, France
- Keio University, Tokyo, Japan
- RIKEN Genomic Sciences Centre, Saitama, Japan
- University of Washington Genome Centre, Seattle, WA, USA
- Gesellschaft für Biotechnologische Forschung mbH, Braunschweig, Germany
- Washington University Genome Sequencing Centre, St. Louis, MO, USA
- Institute for Molecular Biotechnology, Jena, Germany

\textsuperscript{76} Supra note 67.
\textsuperscript{78} Id., at 279.
Potential Benefits of Mapping of Human Genome

As the Human Genome Project reaches completion, the ancient proverb, "knowledge is power," seems truer and more appropriate. DNA underlies almost every aspect of human health, both in function and dysfunction, thereby acting as the controlling mechanism. The knowledge gathered out of HGP is expected to revolutionise medical field. Obtaining a detailed picture of how genes and other DNA sequences work together and interact with environmental factors ultimately will lead to the discovery of pathways involved in normal processes and in disease pathogenesis. Such knowledge will have a profound impact on the way disorders are diagnosed, treated, and prevented and will bring about revolutionary changes in clinical and public health practice. Some of these transformative developments are described below.

79<http://www.genome.gov/11006939> the site is maintained by National human Genome research Institute, National Institutes of Health, Department of Health and Human Services and office of Science U.S department of Energy (last visited on 23-12-2007).
Diagnosis and Prediction of Genetic Disorders

Technology and resources promoted by the Human Genome Project is expected to have profound impacts on biomedical research and promise to revolutionize biological research and clinical medicine. Every person has an estimated five to thirty serious “misspellings” or alterations, in his or her DNA, which means that every individual may be subject to some form of genetic disorders. Increasingly detailed genome maps have aided researchers seeking genes associated with dozens of genetic conditions, to correlate specific genes and chromosomes for approximately 1,050 genetic disorders including myotonic dystrophy, fragile X syndrome, neurofibromatosis types 1 and 2, a kind of inherited colon cancer, Alzheimer’s disease, and familial breast cancer. DNA-based tests are among the first commercial medical applications of the new genetic discoveries. Gene tests can be used to diagnose and confirm disease, even in asymptomatic individuals; provide prognostic information about the course of disease; and, with varying degrees of accuracy, predict the risk of future disease in healthy individuals or their progeny.

Therapeutic Cure for Genetic Disorders

On the horizon is a new era of molecular medicine characterized less by treating symptoms and more by looking to the most fundamental causes of disease. Rapid and more specific diagnostic tests will make possible earlier treatment of countless maladies. Medical researchers also will be able to devise novel therapeutic regimens based on new classes of drugs, immunotherapy techniques, and avoidance of environmental conditions that may trigger disease, and possible augmentation or even replacement

80 Supra note 5, See Jennifer at 211.
81 <http://genome.rtc.riken.go.jp/hgmis/publicat/97pr/02intro.html> (last visited on 13-12-2007)
of defective genes through gene therapy. 83 This largely experimental field—gene transfer or gene therapy—holds potential for treating or even curing such genetic and acquired diseases as cancers and AIDS by using normal genes to supplement or replace defective genes or to bolster a normal function such as immunity.84

There are currently two types of therapies available for such genetic disorders, somatic and germ line, both of which can be developed in response to genes that contribute to disorders. Somatic cell therapy allows for the alteration of cells that make up a person's tissues and organs, known as somatic cells, and results in changes to only that individual. Germ line manipulation replaces missing or defective genes with perfect normal copies of the same genes by inserting them into the sex cells of the patient or the undeveloped cells of an early embryo that is fertilized in vitro.85

Within the next decade, researchers will begin to correlate DNA variants with individual responses to medical treatments, identify particular subgroups of patients, and develop drugs customized for those populations. The discipline that blends pharmacology with genomic capabilities is called pharmacogenomics. 86 More than 100,000 people die each year from adverse responses to medications that may be beneficial to others. Another 2.2 million experience serious reactions, while others fail to respond at all. Future advances will enable rapid testing to determine the patient’s genotype and guide treatment with the most effective drugs, in addition to drastically reducing adverse reactions.87

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84 Supra note 82.
86 Supra note 82.
87 Ibid.
Genomic data and technologies also are expected to make drug development faster, cheaper, and more effective. Most drugs today are based on about 500 molecular targets, but genomic knowledge of genes involved in diseases, disease pathways, and drug-response sites will lead to the discovery of thousands of additional targets. New drugs, aimed at specific sites in the body and at particular biochemical events in the body.88

**Insight into Biology**

"Genomics has come of age, and it is opening the door to entirely new approaches to biology sums up noted scientist Smith"89 The genome research has sparked a change in research paradigm in biology. Famous scientist Smith observed "Some years ago, the central idea or dogma in molecular biology research was that information in DNA directs RNA, and RNA directs proteins. Today, there is a new paradigm to guide us: Sequence implies structure, and structure implies function. The word 'implies' in our new paradigm means there are rules," 90

"Comparative genomics," many believe that whole genomic sequences of different microbes, will probably give the science different ideas about relationships among archaebacteria, eukaryotes, and prokaryotes.91 The growing research in genome will provide a deeper and more comprehensive understanding of the molecular processes underlying life and will have an enduring and profound impact on how we view our own place in it. This may also bring a new perspective to the question of evolution. For instance scientists may study evolution through germ line mutations in lineages besides studying migrations of populations through maternal genetic inheritance. Thus genome sequences are important for

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88 Ibid.
90 Ibid.
91 Ibid.
addressing questions concerning evolutionary biology, the reconstruction of life on this planet, the definition of gene families and the search for a universal ancestor.92

Apart from these potential applications of genome research impact will be expected to leave its mark on molecular medicine, waste control and environmental cleanup, biotechnology, energy sources, and risk assessment.

**Microbial Genomes.**

In 1994, taking advantage of new capabilities developed by the genome project, DOE formulated the Microbial Genome Initiative to sequence the genomes of bacteria useful in the areas of energy production, environmental remediation,93 toxic waste reduction, and industrial processing. Under this project around six microbes thriving under extreme conditions have been sequenced. Further studies are on regarding the uniqueness of proteins of these organisms with the aim to use these microbes and their enzymes for such practical purposes as waste control and environmental cleanup.

**Biotechnology**

Sequencing the genome of other organisms also occupied a prominent place in the goals of HGP. This is expected to bring a host of potential for biotechnology area and industry. The Genome Project already has encouraged momentous investment by multinationals especially in developed countries to capitalize on these potential.94 For instance in the area of energy sources the research to find will play a role in improving the...

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use of fossil-based resources. The energy shortage which will grip the world requires measure and strategies. Biotechnology promises to help address these needs by providing cleaner means for the bioconversion of raw materials to refined products. In addition, there is the possibility of developing entirely new biomass-based energy sources. Having the genomic sequence of the methane-producing micro-organism Methanococcus jannaschii, for example, will enable researchers to explore the process of methanogenesis in more detail and could lead to cheaper production of fuel-grade methane.95

Risk Assessment.

Understanding the human genome will have an enormous impact on the ability to assess risks posed to individuals by environmental exposure to toxic agents. Scientists know that genetic differences make some people more susceptible and others more resistant to such agents. Far more work must be done to determine the genetic basis of such variability. This knowledge will directly address DOE’s long-term mission to understand the effects of low-level exposures to radiation and other energy-related agents, especially in terms of cancer risk.96

Potential Ramifications

New developments in the Human Genome Project and genetic research will make it possible to identify the genetic basis for human diseases, the result will be the invention and making of personalized drugs and individual strategies and method for prevention, detection and treatment. While these developments offer a host of positive result by offering advanced treatment options they could also be put to other ends. Critics of genetic research long ago cautioned, that “[t]he mapping of the genome.

96 Supra note 82.
does not tell us anything about function, which is what the gene sequences do in the organism," and therefore have raised several ethical and legal questions with significant long term implications for humanity. The ethical and moral problems that the HGP faces are probably the most complex and important issues that will ever be addressed by the world.

**Ethical Issues**

One might be tempted to believe that this genetic blueprint might be the closest mankind has come to grasping the Holy Grail. At the same time, from the very beginning of the Human Genome Project, many from both the scientific and public sector have been concerned with ethical issues raised by the research. The ethical issues raised by the human genome project can be grouped under into two general categories: relating to genetic testing and genetic engineering.

The first category consists of ethical questions pertaining to the acquisition and use of genetic information through genetic testing. "Genetic information is not only of value to the individual patient; but employers, insurers, educational institutions, law enforcement officials and others may wish to gain access to an individual's personal genetic profile . . . with a great potential for third parties to misuse information. Once we pinpoint the genetic basis for diseases and other phenotypic traits, through genetic testing the question arises what parameters should be set for the acquisition and use of genetic information? There are the lingering questions relating to fairness in evaluation of genetic facts, consent before and after the test regarding the conduct of test and use of data gathered from the test. Further ethical issues includes preserving the confidentiality of an individual's DNA information and avoiding the stigmatization of individuals who carry certain genes. For instance question arises, Should

genetic information be provided to family members who did not seek the information for themselves but who may be at risk of developing. The question also raises regarding the accessibility of information by social agents like insurers and employers. Some fear that insurers will deny coverage for "pre-existing" conditions to people carrying a gene that predisposes them to particular diseases or those employers might start demanding genetic testing of job applicants. There are fears of exploitation of rare genes in individuals, conduct in reporting results in population genetics, genetic testing of modified DNA on individuals among others.

There are other concerns also regarding genetic testing, one of the scientific limitation of the tests are that the tests may not detect every mutation associated with a particular condition and the ones they do detect may present different risks to various people and populations. Another important consideration in gene testing is the lack of effective treatments or preventive measures for many diseases and conditions now being diagnosed or predicted which will bring anxiety and worries in individual. However the very existence of testing availability gives rise to the following questions: should society ever impose testing on people who are at high risk of developing a condition? Specific type of testing may bring associated problems For Instance, prenatal genetic testing could lead to genetic manipulation or a decision to abort based on undesirable traits disclosed by the tests. In addition, some raise concerns that a full knowledge of the human genome could raise profound psychological issues. For example, individuals who know that they carry detrimental genes may find the knowledge to be too great a burden to bear. All of

99 Ibid.
these ethical issues will ultimately have to be addressed by society as a whole.\textsuperscript{100}

The second category of genetic engineering through somatic and embryonic therapy brings along a host of problems. If current trends are anything to go by, the technologies of the not-so-distant future might well enable mankind to tinker with DNA to the extent of playing God. Humanity has to face the situation of being responsible for tasks, which will increasingly put his own survival as a species at risk. There will be questions of how experimentation in correcting genetic defects should be carried out and with what priority.

Legal Issues

The HGP and the subsequent genetic revolution has changed and is changing our understanding of human identity and raises various challenges to existing legal and social paradigms. Some of the obvious legal issues are: privacy, discrimination, costs, and criminality. While genetic advances will clearly spawn some new issues, others exist already in other contexts.

One of the prominent ethical and legal questions arising out of genetic revolution will be Fairness in access to advanced genetic technologies. The major question in this context will be that who will benefit and will there be major inequities, Looking at the inequities existing in all other fields in society, the inequities in access to genetic technologies is rather a logical conclusion. Even then this is a legal issue worth pondering for the policy makers.

With the success of the HGP, we have overcome the psychological barrier of cracking nature’s code Information about a person’s genetic make-up. This will make it possible to predict his or her health and perhaps even his

\textsuperscript{100} Supra note 7, See Robert at 1187.
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or her behaviour to some extent. The proliferation of information from the Human Genome Project (HGP) is refocusing the debate between an individual’s interest in keeping personal genetic information private and the interests of third parties in gaining access to such information. The question arises who owns and controls genetic information? Is genetic privacy different from medical privacy? Revealing information about the risk of future disease can have significant emotional and psychological effects as well. Additionally, because genetic tests reveal information about individuals and their families, test results can affect family dynamics. Results also can pose risks for population groups if they lead to group stigmatization.101

Further questions arises is it necessary to regulate access to personal genetic information is only on the basis of informed consent. Since such information is likely to be of interest to third parties such as employers and insurance companies. They may want to base their decisions on genetic information, and, in some cases, they might even require employees and or policyholders to undergo genetic testing. What weight age should be given to an individual’s right to know or not to know details of his or her personal genetic information.102

For instance, employers may use genetic information to make employment decisions. The ever increasing panoply of genetic tests gives employers a powerful tool to assess the long term costs of hiring or retaining a particular employee. Employers may opt to deny employment opportunities to certain individuals because they wish to avoid the financial consequences of hiring those who are genetically predisposed to conditions which may make them more likely to take sick leave, resign, retire early for health reasons, file for workers’ compensation, or use

101 Supra note 82.

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health care benefits excessively. Genetic testing could thus become a
definitive factor for employers in deciding whom to hire thus leading to
genetic Discrimination.\textsuperscript{103}

The same scenario can be repeated in Insurance field where Insurers may
decide to insure or charge high premium from people having genetic
defects. The misuse of personal genetic information and consequent
discrimination can be expected to find its way to education field adoption
field, Immigration etc. Most of the experts fear that access to genetic
information may give rise to discrimination, and that individuals need legal
protection against such discrimination.\textsuperscript{104}

Genetic advances also promise to give rise to new issues in the field of
criminal law. In most criminal cases, the prosecution must prove that the
accused both committed and intended to commit the wrongful act.
Suppose a gene is discovered that predisposes an individual to violent,
predatory behaviour. Will an individual possessing such a trait be entitled
to plead that his genes made him do it if he is charged with a criminal
offence? In other words, will courts acquit an individual who admits a
criminal act but denies responsibility as he had no control over his
genetically programmed impulses? In such a scenario, the individual may
have intended his actions. However this intention may have arisen from
his genetic makeup and not his own free will. Will this allow an accused to
successfully deny that he has had a guilty intention? Or will the courts find
that the intent element of the offence is made out notwithstanding that the
accused could not help the intention?\textsuperscript{105}

Genetic science and related technologies are important in medical
research and in the development and provision of healthcare, and, their

\textsuperscript{103} Id., at 641.
105 Maureen P. Coffey, Note, "The Genetic Defence: Excuse or Explanation", 35 Wm. &
significance for human health is likely to increase as more becomes known about the biological functions of genes and the proteins they produce. Human genetic research may translate into the development and provision of new forms of healthcare involving, among other things, medical genetic testing, pharmacogenetics, gene therapy, and the use of therapeutic proteins or stem cells. There are many ways in which the potential subject matter of these technologies may give rise to the issue of gene patenting. The question arises whether it is ethical and legal to allow ownership or patent to something which is the common heritage of humanity. Further questions arises, Will patenting DNA sequences limit their accessibility and development into useful products? All these issues need to be addressed by society as a whole.

Lastly but not the least With advances in genetic science that make it possible to identify genes that are known to make a person susceptible to a particular disease, a prophylactic treatment may be developed that, if administered before the onset of the disease, may prevent it entirely. Should insurance companies, Medicare or Medicaid be required to cover expensive genetic testing and prophylactic treatments? While such testing and treatments provide obvious benefits to both the individual and society, the cost could be prohibitive, especially as more and more genes are identified that could prompt testing and treatment.

**Concluding Remarks**

The decoding of the complete genome sequence has been said to be the greatest adventure in modern science, and its success has been rightly called as the end of the beginning. It is likely to open many new areas in

106 Supra note 75.
human biomedical research. There is definitely a new ray of hope for millions of people who suffer from diseases whose underlying causes are yet to be understood at the base level. Although much needs to be done in many of these cases, the genome sequence provides a fertile hunting ground for searching for the culprit genes. Meaning although the raw text is in place and in the correct order, the grammar and semantics has still to be worked out which will ultimately reveal. In his classic novel George Orwell wrote “All history was a palimpsest, scraped clean and re inscribed exactly as often as necessary”. The genome sequence can be considered as a genetic palimpsest, a text that has been overwritten, time and again by evolution, Buried in this sequence, once scientists have developed sufficiently sophisticated tools and computer programs to unearth them, are the answers to the origins of life, the evolution of humanity, and the future of medicine. 108

108 Supra note 17, See Kevin at 251.