H

Human Genome Project

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Synonyms

HGP

Definition

The Human Genome Project (HGP) is an international, interdisciplinary, scientific research project aimed at determining the sequence of chemical base pairs which make up human DNA, mapping the entire human genome, and identifying its complex structures and functions.

Introduction

The Human Genome Project (HGP) is an international, interdisciplinary, scientific research project aimed at determining the sequence of chemical base pairs which make up human DNA, mapping the entire human genome, and identifying its complex structures and functions. The outcomes of the project have been established as open and accessible to all. Leading healthcare, research, and disease advocacy organizations from more than 40 countries coordinated a global alliance for genetic health dedicated to enabling the secure sharing of genomic and clinical data in a standardized (technical and regulatory), effective, and responsible manner. Sequencing the human genome in its entirety substantially furthers our understanding of human evolution, the causes and mechanisms of disease, and the complex interactions between genes and environment. Challenges over the next decade include interpreting the contents of all the sequenced genes and understanding how they function. Genome-based research will eventually enable medical science to develop highly effective diagnostic tools, to better understand health needs based on individual genetic make-ups, and to design new, personalized treatments for disease.

History

The Human Genome Project (HGP) was first publically advocated in 1984 by Renato Dulbecco. In 1985, a dozen experts convened in Santa Cruz, California, to discuss the feasibility and professional implications of the HGP and concluded that it would be possible, but challenging. The HGP was controversial among these experts. Those opposing the HGP argued that sequencing the human genome was not worthwhile because it is mostly junk, which current technology was not up to the task, and that manually sequencing the genome was too mundane of a job to be attractive

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to talented people. The HGP continued to be objected to throughout the 1980s by the majority of biologists, as well as the National Institutes of Health. Crucial support for the HGP came from the US Department of Energy in 1986, which supported the project because they sought data on protecting the genome from gene-mutating effects of radiation. Consequently, an early genome project was established in 1987 (National Human Genome Research Institute – NHGRI 2012) under the direction of the National Institutes of Health and the US Department of Energy, entailing a 15-year, $3 billion, plan to complete the human genome sequence. Parallel to the US government-sponsored HGP, the American researcher Craig Venter, through his firm Celera Genomics, announced in 1998 his intention to build a unique genome-sequencing facility to determine the sequence of the human genome over a 3-year period.

The first analyses of the draft human genome sequence were reported in the February 2001 issues of *Science* and *Nature*. The *Nature* publications included initial sequence analyses generated by the publicly sponsored HGP (see Lander et al. 2001), while the *Science* publications focused on the draft sequence reported by the private company Celera Genomics (see Venter et al. 2001). In 2003, the HGP was declared complete (Human Genome Project Information Archive – HGPIA 2015).The studies announcing completion reported 99% of the euchromatic human genome with 99.99% accuracy (International Human Genome Sequencing Consortium – IHGSC 2004), followed by a major quality assessment of the human genome sequence that indicated over 92% of sampling exceeded 99.99% accuracy (Schmutz et al. 2004). At the time, the human genome was estimated to contain approximately 20,000–25,000 genes.

The main differences between the draft (Lander et al. 2001; Venter et al. 2001) and finished versions of the human genome sequence (IHGSC 2004) were the percentages of genome covered, the number of gaps, and the error rates. The draft sequence covered 90% of the genome at an error rate of 1 in 1,000 base pairs, and there were over 150,000 gaps, with only 28% of the genome reaching finished standard. In the finished version, there were less than 400 gaps, and 99% of the genome was sequenced with an error rate of less than 1 in 10,000 base pairs. These differences are significant for scientists using the sequence to conduct research (NHGRI 2010). After the human genome sequencing was complete, the US Department of Justice filed a court brief stating that genes should not be eligible for patents because they are products of nature. Thus, the human genome database is publicly available to anyone (see The Genome Database – GDB, gdb.org).

Analyses of the HGP data are ongoing (see HGPIA 2015). For instance, in 2012, the Encyclopedia of DNA Elements (i.e., ENCODE) published results from a cross-consortium integrative analysis, covering more than four million regulatory regions in the human genome in 30 coordinated papers in *Nature*, *Science*, and other journals (see encodeproject.org). The ENCODE website allows readers to follow a topic through all the papers in a publication set. In 2013, over 70 leading healthcare, research, and disease advocacy organizations from more than 40 countries coordinated a global alliance for genetic health dedicated to enabling the secure sharing of genomic and clinical data in a standardized (technical and regulatory), effective, and responsible manner (see http://web.ornl.gov/sci/techresources/Human_Genome).

Applications

Sequencing the human genome in its entirety substantially furthers our understanding of human evolution, the causes and mechanisms of disease, and the complex interactions between genes and environment. The HGP importantly contributes to health improvement in many ways. For example, individualized analysis based on a person's unique DNA has the potential to be a powerful tool for medical prevention and treatment (NHGRI 2010). Physicians will be able to better predict future risks of illness onset and potentially

harmful behaviors that impact each individual. Nurses, genetic counselors, and other healthcare professionals will be able to focus their efforts on aspects that are most likely to maintain or improve individual patients' health. These aspects may include personalized dietary and lifestyle changes and targeted medical monitoring. Understanding of prevalent diseases such as diabetes, heart disease, and schizophrenia at the genetic and molecular level may revolutionize healthcare through earlier, more precise, detection and, therefore, intervention.

Application of new and more effective drugs based on the completed genome sequencing are at least 10–15 years in the future, although more than 350 biotech products – many based on the HGP – are currently in clinical trials (NHGRI 2010). Usually, it takes over a decade for companies to conduct the clinical studies that are needed for marketing approval from reputable institutions. Testing for health risks and genetic predispositions to disease, however, will arrive more quickly – in particular abilities to predict individual future health risks and implement an enhanced approach to preventive medicine. Moreover, researchers and physicians will be able to better determine which drugs will provide the best outcomes for particular individuals, based on their genetic make-up (NHGRI 2010).

One of the largest challenges over the next decade will be interpreting the contents of all the sequenced genes and understanding how they function – including their role in human health and pathology (NHGRI 2010). Genome-based research will eventually enable medical science to develop highly effective diagnostic tools, to better understand health needs based on individual genetic make-ups, and to design new, personalized treatments for disease.

Future Directions

The HGP and consequent deeper knowledge of our genome has revolutionized the practice of medicine, inspiring several large-scale data acquisition initiatives such as the 1000 Genomes Project, the Chimpanzee Genome Project, the Neanderthal Genome Project, and the Cancer Genome Atlas. Starting in 2008, an international research initiative called the 1000 Genomes Project set out to create a complete and detailed catalogue of human genetic variations (www.1000genomes.org). Such a catalogue will be useful in association studies that link genetic variation to disease. Another related project is the Chimpanzee Genome Project, which was created in 2003 and aimed to determine the DNA sequence of the chimpanzee genome. Comparing the genomes of humans and other apes will shed new light on what makes humans genetically distinct from, and similar to, other species. Similarly, the Neanderthal Genome Project is an endeavor to sequence the Neanderthal genome (www.neanderthal.ensemblgenomes.org). The Neanderthal Genome Project published their results in 2010 in *Science*, detailing an initial draft of the Neanderthal genome based on the analysis of four billion base pairs of Neanderthal DNA. Other projects inspired by the HGP include the Human Brain Project and the emerging Human Proteome Project, both of which were recently launched and hold great promise for the future of medicine and psychology (reviewed in Hood and Rowen 2013).

Conclusion

The HGP has transformed biology through its interdisciplinary approach to deciphering a reference human genome sequence. The HGP exemplifies the power, necessity, and success of large, integrated, cross-disciplinary efforts (e.g., engineering, computer science, math, and biology) directed toward complex major objectives. This ambitious endeavor led to the development of novel technologies and analytical tools (reviewed in Hood and Rowen 2013). Importantly, the outcomes of the project have been established as open and accessible to all. Finally, the HGP has inspired several other exciting projects that promise to open new avenues in biology, medicine, and psychology.

Cross-References

- ▶ Genetic Basis of Traits
- ▶ Genetic Diversification
- ▶ Genetic Variation
- ▶ Molecular Genetics

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