Editorial

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Human genome at 20: from an extensive international effort to a wealth of discovery

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The human genome project (HGP) symbolises one of the most audacious, extensive and successful collaborative research feats of the last century. The HGP was an aspirational venture to decode the DNA sequence of the whole human genome. The proposal met with much censure. Some people assumed that the venture was too gigantic and ambitious to get ahead; others counter it as a biological philately. Disbelievers doubted that sequencing the human genome would be helpful and adjudged it a profitless activity and tremendous waste of money. After ten years – with the expense of billions of dollars, uncountable hours of efforts on DNA sequencing, and an enormous travail from an international collaboration of twenty institutions from six nations – the preliminary draft of the Human Genome was brought out in June 2000 with an extravagant function at the White House and massive media attention. The project officially announced its goal in 2003 with the publication of an updated reference genome.

The release of the first draft of a 2.91-billion base-pair consensus sequence of the human genome in 2001 was a landmark achievement [1, 2]. Decoding the DNA that makes up the human genome has been extensively expected for its contribution to interpreting human evolution, the cause of disease, and the interaction between the environment and inheritance in delineating the human condition. It was expected that having excess to the genome sequence would render and transform our understanding of the human disease leading to better treatment detection and prevention.

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With the completion of the human gene map in 2003, we gained more detailed information on the structure and function of the vast genes involved in disease susceptibility. We developed gene therapy strategies with the application of a molecular approach to treat such diseases. Its achievement should be evaluated by how this venture changed the directives of research, practising biological breakthrough, and the universal digitisation of natural science - the benefits of an entire cancer genome, developments in genetics, and bettered technologies [3]. Celebrating 20 years, we should also reflect on what the 20 years in a post-genome world have wrought (Figure 1) and what to expect in the next 20 years. This article will make you see through the HGP from its beginning to the released sequence and into the completely developed arena of human genomics. Today, the HGP became a luminary in the domain of genes, genomes and biomedical genomics.

During the inception of the HGP in 1990, it was expected that the HGP data would become the reference for biomedical science in the 21st century and cause the healthcare revolution [2]. Remarkably, there would be information on all genes, disorders, and genetic variations. Clinical geneticists recognised that this would decipher and dissect the molecular basis of hereditary childhood diseases. At the same time, adult disease specialists looked for solutions to some plebeian ailments such as heart diseases or cancer. Technologists recognised that this project would be the entrance to the new age of high-throughput, digital biology. The culmination of the HGP in 2003 marked genome research as a cardinal scaffolding to biomedical science. The outcome of HGP has been paving the means for a health care evolution, established on the groundwork of the improved understanding of the molecular basis of human disease pathogenesis. Novel and advancing technologies in computational biology and bioinformatics will permit high-throughput, sophisticated gene-based diagnostics that will forever change biology and healthcare practice. The flagship endeavour of the HGP was to decode the three billion base pairs in the human genome, the physical assembly of the DNA, and the position of the predicted 30,000 genes. Similar to this effort, the DNA of model organisms was sequenced to offer the comparative data essential for interpreting the human genome's function. And here we are today, two decades after that inaugural announcement, with several thousands of sequenced human genomes, bringing out comprehensive knowledge about

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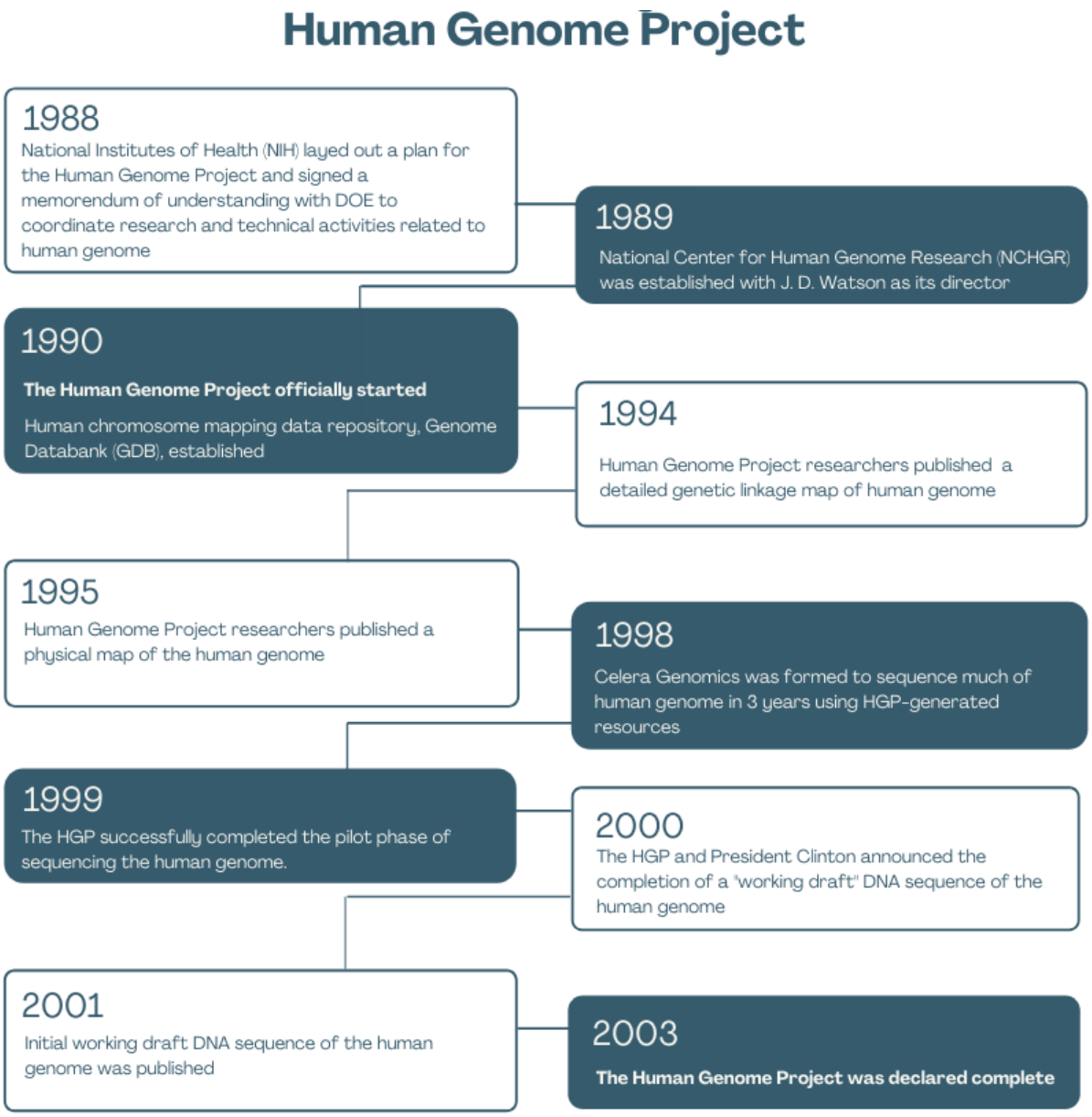

Figure 1: The timeline of the Human Genome Project

health, disease and human biology, with the reimbursements of genomics opening to percolate into mainstream medicine.

The origin of the HGP is the basis of genomics, the word that did not exist until 1987; that's when the term 'genomics' was coined, and it was initially used in the scientific literature in a very recent academic journal of that time called genomics. And you could raise a question why in the late 1980s, was there a necessity to identify a new area of study and anticipate a big international bodacious venture that ultimately turns out to be the Human Genome Project. It followed out various advancements that transpire over decades, in fact of the preceding century, beginning with the Watson-Crick's double-helical model of DNA in the 1950s, the elucidation of the genetic code in the 1960s. In the late 1970s and early 1980s, when molecular biology tools took hold, our approaches for isolating, cloning and manipulating DNA, and most highly, reading DNA for essentially sequencing DNA. All of those tools and technologies are currently having extensive utility throughout the biomedical research landscape. The genome project generated an almost complete reference sequence of the human genome (99%), but the technology forbids it to make it entirely complete.

As the draft sequence of the human genome was declared, instantly, the intent shifted towards writing up the papers that the focus would be on concluding a high-quality sequence. When the HGP concluded, it brings about keen enthusiasm about all that would follow, a broad recognition that the best was still to come. In several ways, that was just the beginning.

But then again, even single representation of the human genome is insufficient. A single human genome sequence can't characterise even an individual since we all have two sets of the human genome in ourselves; one from the mother and another from the father. So, the ultimate challenge now is how to present the human genome sequence and indicate

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the variation, at least the typical ones, throughout humankind all around the globe. Both experimental and computational technologies are being developed to signify the diverseness of genome sequences distinct by our species. Besides the conception of the human genome to the global genome, people are now working on various other aspects involving the genes, such as the cancer genome, and delving into specific tissues and genetic variations and all these types of things. The way the technology is enabling us to capture this diverseness and variance is simply unbelievable. And it is imperative to comprehend how we correspond various people and the alteration that occurs and how that varies or may explain the differences in our characteristics. But it is crucial to analyse the individual's genomic data with the relevance to healthcare, whether the treatment is possibly relevant for them or not, and this is referred to as precision medicine.

It's been twenty years since the HGP brought out a draft of human genetic reference, which assured that doctors would soon be looking at a person's DNA and recommend the precise medications for that person's illness or even thwart certain diseases. That promise has thus far to be accomplished in any prevalent way. Investigators are acquiring hints about genetic variants associated with certain conditions and how medicines act upon the target. But numerous such developments have helped people belonging to Europe. And so, there are also medical grounds for the requirement of these very superior-quality reference sequences from individuals of diverse ancestral assemblages.

It is imperative to emphasise the difference between where we are today. Even when the HGP ended 18 years ago, the arena of genomics was titular and very limited. By 2011, when the second strategic vision of HGP was published, it began to propagate, but today, one and all are doing genomics. And so, it's practically inconceivable to do strategically contrive around encompassing genomics.

And it is fascinating, and it is throughout the entire range, upheld significant vehemence on apprehending the fundamental structure and function of the human genome. Pushing these genomic understandings into the clinical realm and applying genomics in healthcare and medicine is being realised over the past ten years, but that's only initial instances. As we know, healthcare is very intricate in almost every single country, and so genomics has exceptional potency. Yet, it has to make its way into the composite ecosystem of healthcare deliverance and become more dynamic in medicine over time. It deciphers entire operational elements, discerning how variation determines function and then actuating into translational prospects of variation in human health and disease. Looking into the genomic architecture of human diseases would evaluate the success of genomics-driven healthcare and medicine.

It is an entirely antithetic world today in 2021, compared to 1990. All experimental biologists and geneticists are now mandatorily required to be trained in computational biology and have easy access to massif principal and consequential data to be anticipated. With the emergence of the recent COVID-19 pandemic, several researchers and scholars are coerced to stay away from the wet lab, swivelled to computational studies; about 30-35 years ago, they would have vanished. The actual upshot of the HGP lies in the eminence amid the initial state of digital biology in the late 1980s and the existing affluence by means of which all scholars can access, connect and study biological data.

Several health conditions observed in kids and grown-ups have a solid, prevalent, genetic basis. It is impartial to state that virtually all the human medical conditions are associated with a hereditary influence [4]. Online Mendelian Inheritance in Man (OMIM) is a continuously updated list constituting many thousands of such disorders [5]. However, OMIM offers a way too small perspective on contribution of genetics to medicine [6]. Thanks to the HGP, today, we have a comprehensive genomic map and the repertoire of information regarding all genes, genetic variants and the molecular basis of the Mendelian diseases. But merely having a map won't provide all the necessary info. The project didn't give a detailed account of the activation of those genes to bring out exclusive organs, unique individuals, and inimitable diseases. While the HGP gave us an extraordinary amount of information about our beingness, it was only a footmark in understanding how and why we are what we are.

This is where epigenomics will comprehend how environmental exposures and lifestyle behaviours lead to disorders. By looking at the standard patterns of the epigenome, we can locate the errors or alterations that crusade diseases from cancer to mental ailments. And it will bid novel and exciting targets for medicaments to treat diseases that have epigenetic factors. Hence, genomic data revolutionised healthcare practices. Still, the future of precision medicine is associated with the advancement of epigenomes requiring multiple maps to be generated from many samples of various tissues, cells and situations.

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