ISSN No:-2456-2165

The Revolution of Genomics Technologies and their Applications in Basic and Clinical Science

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Abstract:- The Human Genome Project focuses on identifying and mapping human genome genes and the base pair of DNA. Bioinformatics is an emerging field that plays a significant role in exploiting the existing amount of genome data to solve a wide range of genomic problems.

The emergence and implementation of bioinformatics are vital in providing a platform for managing, analysing, and implementing the existing data on genomics. One of the leading benefits of bioinformatics is the increased capacity to perform system-wide analysis involving genomes, phonemes, and explanations on the association between phenotypes and genotypes. Modern technologies and revolutions in genomics impact a wide range of areas and are beneficial in resolving diverse problems in these fields, including environmental impacts, low-profit margins, and diseases.

The revolution significantly influences third-generation sequencing to overcome second-generation sequencing weaknesses in various genome regions. Genomics' future is unpredictable and is anticipated to accelerate faster, accompanied by security, privacy, and human genomic challenges. Based on the previous impacts, genomics will continue to have immeasurable effects on different fields, the environment, biology, and society. The concepts of epigenomics, functional genomics, and metagenomics reveal the benefits of genomics – a broad area relevant and significant in the current era. Genomic concepts can be used to solve a wide range of challenges, especially concerning third-generation sequencing.

In the current era, genomics is not only beneficial. Modern technology has also significantly revolutionised to enhance the implementation in solving problems with genes and proteins and their interactions.

Keywords:- *"Genomics," "human genome," "bioinformatics," "genomic problems," "emergence and implementation of bioinformatics," "revolution of genomic," "genomic concepts," "third-generation sequencing," "genomic practice."*

I. INTRODUCTION

The field of genomics started being popular towards the end of the twentieth century. The completion of the Human Genome Project particularly played a significant role in the start of the genomics revolution, through the discovery of the whole set of human genes, which would be available for extensive biological research [1]. The Human Genome Project aimed at identifying and mapping of the human genome genes, as well as investigating the base pairs that make up the DNA from both the functional and physical points of view [2]. The human genomics experiments result in genomics and the publishing of the first mouse genome and human sequence, which were major milestones [3]. Essentially, genomics deals with the study of the evolution, organization, structure, structure, expression, and functions of the genome. Genomics relies on large sets of data, which create challenges and demands for new approaches to data management. Most of the data that genomics draws comes from population genetics, quantitative genetics, molecular genetics, and cytogenetics. The concept of bioinformatics is one of the areas that has emerged as a result of the vast existing demands created by the vast amounts of data. Bioinformatics plays a crucial role in solving a diverse range of data needs such as collection, storage, analysis, and integration of data generated from genomic research. Figure 1 below illustrates the key disciplines of bioinformatics.

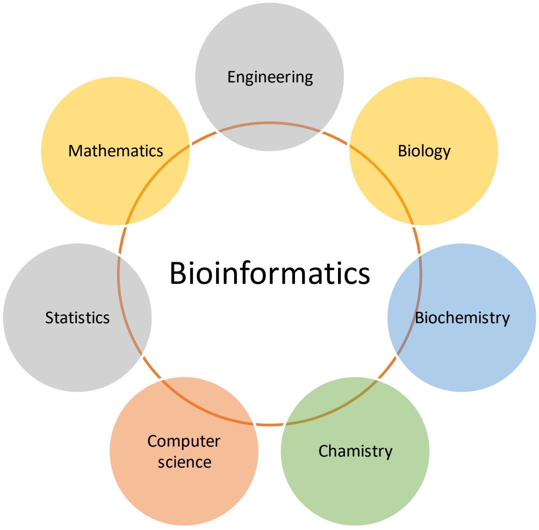

Fig. 1: Key disciplines in bioinformatics

II. RESULTS

The emergence of bioinformatics plays a crucial role in the implementation of genomics in diverse ways by providing a platform for data analysis, data management, and implementation [3]. Specifically, as it can be seen on Figure 1, bioinformatics develops and applied a diverse range of concepts including statistics, mathematics, and other informatics tools are applied in studying and interpreting genomics.

The new scientific discipline makes it possible for the integration of data from several fields, including phenomics, metabolomics, proteomics, and transcriptomics, as well as the use of both genomic and post-genomic data. Metabolomics deals with the study of netabolite profiles while investigations on protein enconded by a genome is proteomics. Trascriptomics deals with gene expression processes [3]. One of the benefits of the integration of the different fields is that it allows system-wide analysis involving the phoneme, genome, and the explanation of functional association that exists between the genotypes and phenotypes [4]. Genomics revolutions and technologies impact a diverse range of areas in resolving problems in different areas such as low-profit margins, environmental impacts, and diseases, as it is revealed in the study by Liu [5].

Studies have documented various aspects of the past, present, and future genomics technology. Dr. Tom Roderick coined the term "genomics" in 1986, which happened in a meeting in Maryland on the topic of the human genome. The term genome was developed from "Genom," a German word. Crockett and Crockett defines genomics as an area in biology that focusses on the evolution, function, structure, mapping, and editing of the genomes [6]. As Ogbe et al. cite, it is until 1926 that the term was used in English [4]. At this time, as Ankey (2013) indicates, there were still unique efforts applied to gene sequencing. Key individuals at this point in the field of genomics included Francis Crick and Fred Sanger, who had done tremendous work on the insulin's amino acid sequence. Earlier on in 1941, Franklin had described the existence of the helical structure in DNA (Ankey, 2013). An empirical study by Holley et al. provided valuable insights into the works of genomics in the early years. Robert W. Holley, in collaboration with several other researchers, produced the first publication on nucleic acid sequence [7]. The works by Philip Leder and Marshall Nirenberg were also major contributions to the field of genomics by performing experiments to explicate the genetic code's triplet nature [8].

In the current era, modern technology has revolutionized the practices of genomics immensely [4]. Rhoads and Au espouse the role of technology in the current practices of genomics, including research. The investigators reveal that high-sequencing throughput technologies have created a platform for third-generation sequencing [9]. One of the advantages of third-generation sequencing is that it overcomes the weaknesses of second-generation sequencing, which include that reads that were generated were short and could equal various regions of the genome [10]. Ye et al. also confirm that the ability of the third generation reads to generate long erroneous reads is a key milestone that has been achieved in the field of genomics [11].

Advantages	**Disadvantages**
Can detect anomalies occurring in the entire genome sequence	Lack of computational resources ion many organisations may hinder the usefulness of third-generation sequencing in many organisations.
Less DNA is required in detecting the duplications, insertions, deletions, and substitutions.	Currently, clinical significance of the abnormalities is unknown
Greatly reduced time and costs in conducting next generation sequencing.	Requires large data storage capacities, fast data processing, and sophisticated bioinformatics systems.

Table 1: Advantages and disadvantages of third-generation sequencing

Liu describes several changes that have happened in the field of genomics [5]. One of the study by Cheifet provides perspectives of many researchers about the future of genomics, for instance, Cheifet cites Mihaela Zavolan of the University of Basel, who identifies that the future of genomics will entail large amount of data with high resolution [12]. According to Liu describes that one of the areas of genomics is it going functional [5]. Three areas of functional genomics are the application of genomics to biology, genomics to health, and genomics to society. Yu et al. also agree that the emergence of functional genomics is another key feature of genomics in the current era [13]. The emergence of functional genomics can be attributed to advances in modern technology. Functional genomics deals with attempts to explain the working of genes and proteins, as well as the types of interactions that occur. Functional genomics relies on a vast amount of data collected for RNA sequencing, genome sequencing projects, and other types of transcriptomic and genomic projects, such as RNA sequencing and genome sequencing projects [5]. Some of the areas of focus of functional genomics include protein-protein interactions, regulation of gene expression, translation, and gene transcription. As Suter et al. indicate, functional genomic technologies focus on a diverse range of dynamics, which include protein-protein interactions, regulation of gene expression, as well as translation and gene transcription, but not DNA sequences, structures, and other genetic information that are static [14]. Moreover, other key features of functional genomics studies, as Hornung, Zwittink, and Kuijper indicate, apply high throughput methods and other genome-wide approaches as opposed to geneby-gene techniques that are rather traditional [15]. Other areas of genomics in the modern era include the areas of predictive biology, systems biology, and metagenomics. Figure 2 below summarizes the processes involved in functional genomics:

Fig. 2: Functional genomics process

Figure 2 highlights the processes of functional genomics including conducting biological experiments, high throughput experiments, bioinformatics analysis and results. The different stages gives an illustration of changes in genomics. The current practices on genomics face a lot of challenges due to the challenges related to ethics. A lot of controversies and ethical issues surrounding genomics technology exist. As Goodrich et al. there is a diverse range of medical, research, legal, and ethical challenges, especially concerning genomic industries [16]. McGowan et al. also reiterate that although genomics technologies have increasingly become popular due to the impact they have on preventing adverse health outcomes, predicting appropriate therapies, and provision of individualized health care, ethical and social implications are still major concerns [17]. Moreover, McGowan et al. show that ethical concerns about genomics are specific to given fields [17]. The World Health Organization (WHO) also includes ethical and legal concerns as one of the major issues that genomics technology faces. However, according to WHO, the question of DNA patents and the flexibility in international frameworks also needs to be addressed for the full potential of genomics to be realized.

The future of genomics is expected to accelerate faster, but with the challenges of privacy and security, especially in human genomics expected to persist [17]. Also, as Rougemont et al. shows, significant discoveries in genomics will be made, but some are likely to have unintentional or adverse impacts [18]. The outcomes of the human genomics project show that there is a lot of potential for new develops in the field of genomics, especially with the advances in sequencing technology. Twenty years ago, single genome sequencing was impossible but sequencing several thousands of human genome is possible at the current moment [19]. This trend reveals that the capacities, efficiencies, as well as the ability increased abilities for genomics data analysis are great indicators of significant advances in genomics technology. Liu also cautions that some of complex genomics data analysis, which is computer-mediated, are likely to result in some discoveries that deviate from the initial purpose [5].

III. DISCUSSION

After gaining a lot of into the past, current, and future aspects of the technology, I am convinced that genomics will continually have an insurmountable impact on biology, the environment, and society. Before this course, there are several areas of genomics that I was not well-versed with. For instance, I only had a limited understanding of such concepts as metagenomics, epigenomics, and functional genomics. However, with the completion of the course, I now appreciate that genomics is a rich and broad field that has tremendously attained significance in the current era. Also, through this course, I understand the great potential of genomics technology in the current era. As Schadt, Turner, and Kasarskis indicate, there is a great potential of genomics solving a diverse range of challenges in the current era, especially with the advent of third-generation sequencing [20]. The role of third-degree sequencing is an area that I did not have good knowledge about before the start of the course.

Lee et al. mentions that the major advantages of third-generation sequencing as compared to second-generation sequencing is the aspect is longer read, epigenetics, and portability and speed [21]. All these advantages, coupled with the concepts of bioinformatics, reveals that genomics technology has great potential for the future. I have a similar opinion with Cheifet on the question of "where is genomics going next?" Cheifet acknowledges that a significant number of studies about genomics were published on genomics, which is an implication that a lot of research is happening in this field [12]. I anticipate that huge steps will be made in the coming years, especially with the increasing advancement of modern technologies. Also, as Knight et al. indicate, I agree that that the accuracy and efficiency of genomics, which is a key underpinning of metagenomics, will greatly improve [22]. As well, more investments will be channelled to this field, especially by considering that the benefits are bound to outweigh the required investments and potential adverse effects overly.

Genomics technologies are also widely applied in clinical sciences to identify common medical conditions, which include: Diabetes mellitus [27][28][29][30][31][32][33][34], neurodegenerative disorders [35][36][37], Covid-19[38][39][40][41][42], zoonotic diseases [43], cancer [44][45][46][47][48][49][50], degenerative mitral valve diseases [51], genetic disorders [52], metabolic disorders [53][54][55][56].

IV. CONCLUSION

Genomics entails studying the evolution, organisation, structure, expression, and genome functions. Bioinformatics is crucial in solving varied data needs involving existing data from genomic researches. Its benefits are the increased capacity to perform system-wide analysis involving genomes, phoneme, and explanations on the association between phenotypes and genotypes. Nonetheless, the genomic practice is associated with numerous challenges related to ethics and social implications. Its future is unpredictable but anticipated to accelerate faster alongside security, privacy, and human genomic challenges. The relevance and benefits of genomic in the current era are expected to continue outweighing the investment costs or the associated potential risks, legal, security, or ethical concerns.

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