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RESEARCH

Unlocking the potential of personalized medicine - A review

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Bioinformatics is a multidisciplinary field that evolves techniques and software tools for understanding the data in biology, when there are large data sets in particular. This field includes computer science, biology, physics, chemistry, mathematics, and statistics. Recent advancements in high-throughout technologies have been made possible by the emergence of systems biology as a comprehensive science, allowing for a more precise modeling of complicated disorders. A general definition of personalized medicine is a participatory, preventative, personalized, and predictive healthcare strategy. One of the biggest challenges in personalized medicine is the translation of scientific findings into more effective therapeutic results. Translational bioinformatics is a potent technique for bridging the gaps between systems biology research and clinical practice. The objective is to give scientists the appropriate tools for explaining normal biological processes, dysfunctions of these systems that result in disease, and strategies that enable the development of novel therapeutics. Recently, sequencing platforms and the vast amount of genomes and transcriptomes have presented significant problems for genomics in general and bioinformatics in particular. These methods are essential for comprehending disorders and using personalized medicine at systemic levels. The goal of this article is to assemble a list of resources and methods used by scientists to handle data from recent platforms' large sequencing to new generations and the uses of this data in various fields of the life sciences, including medicine.

Keywords: bioinformatics, personalized medicine, translational bioinformatics, next generation sequencing, single nucleotide polymorphism

Introduction

Bioinformatics has been a crucial component of biomedical science research and development over the last 10 years. It is appropriate to reflect on the incredible journey that led to the field's current status when considering a vision for the future of this new section of the biological sciences (1).

Emerging and promising fields like systems biology and pharmacogenomics will provide us a complete understanding of diseases and allow for personalized treatment. In pharmacogenomics, the genetic basis of individual variability in responsiveness to pharmacological interventions is investigated. It aims to achieve personalized medicine by predicting illness susceptibility and medication and vaccination response (2). The term personalized medicine is used to refer genomic medicine. It uses genome knowledge, which is taken from humans and other organisms and its derivatives, which are RNA, proteins, and metabolites, to direct medical decision-making. Future healthcare systems with a more proactive and predictive approach to medicine, where the emphasis is on preventing diseases rather than symptom treatment, are outlined by the concept of personalized medicines (3). The individualization of care for each patient would be the foundation of this method, with all the patient's medical information being available and included computationally. Over the past 10 years, molecular science has contributed to numerous

medical advancements, notably the Human Genome Project and the International HapMap Project (4).

Literature review

A general definition of personalized medicine is a participatory, preventative, customized, and predictive healthcare model. In the upcoming years, the discipline of bioinformatics will be swamped with individual genomic data. The bioinformatics community must address the serious challenges that this data flood raises.

Sarachan et al. in their study, the authors explore the possibilities for creating personalized medicine tools by combining medical informatics—the use of information technology in healthcare and bioinformatics—the use of computer tools for biological data analysis. It probably discusses how combining these 2 professions might enhance patient care and therapeutic results.

Chen et al. in their paper emphasize the use of next-generation sequencing (NGS) technologies in personalized medicine. The development of NGS has transformed genetic research and holds the promise of enabling personalized treatment plans. However, the essay also goes through the difficulties and restrictions that come with integrating NGS into clinical practice.

Rana M. in his paper examines how bioinformatics is influencing the field of personalised medicine. It goes through new developments, techniques, and tools in bioinformatics that aid in developing and using of personalised medicine strategies.

Fernald, G. H. et al. in their paper address the bioinformatics challenges related to personalized medicine. As medical treatment becomes more tailored to individual patients, bioinformatics plays a crucial role in analyzing and interpreting large-scale genomic data to inform clinical decisions. The paper highlights specific challenges in this area and propose potential solutions.

Branco I et al. in their article discuss the most recent developments in bioinformatics tools and how they are being used in the fields of life sciences and personalized medicine. It includes subjects like protein structure prediction, systems biology, and genomic analysis to personalized medicine. The potential of these new tools to advance medical research and enhance patient outcomes are also highlighted in this paper.

Molidor, R. et al. in this paper explore the newest developments in bioinformatics, focusing on the change from genomic sequencing to personalized treatment. It could occur over how to use clinical and genetic data together to customize a patient's medical care. The authors looks at how genetic changes linked to diseases might be identified using bioinformatics methods, informing personalized treatment plans.

Yan, Q. in his paper delves into the field of translational bioinformatics and systems biology as they relate to personalized medicine. The author discusses how computational approaches are used to integrate diverse biological data, such as genomics, proteomics, and clinical information, to gain insights into disease mechanisms and individual patient characteristics. The paper provides examples of how these approaches are applied in drug discovery, treatment selection, and patient stratification for personalized medicine.

In the coming sections, we will be reviewing the method of development and uses of personalized medicines in the field of bioinformatics.

Evolution of disease

Genetic disorder is an essential product of evolution. DNA replication, transcription, and translation are fundamental biological processes that have emerged very early in the history of life. The potential for disease was also generated by these early evolutionary breakthroughs, which gave rise to cellular life. Later developments throughout the long evolutionary history of life have similarly made adaptation and the potential for malfunction possible. Young genetic variations unique to the human lineage combine with contemporary surroundings to form human illness phenotypes against this ancient backdrop. As a result, while the genetic polymorphisms that give rise to genetic diseases in modern people are typically specific to humans, the substrates for these diseases are frequently far older than the human lineage itself.

Millions of people from all around the world are having their genotypes and phenotypes described by massive national biobanks. Our view of the genetic foundation of disease is being fundamentally altered by these studies. The ability to extract and analyze ancient DNA from thousands-of-years-old organisms has also made it possible for researchers to reconstruct the history of recent human adaptation with remarkable detail. These discoveries have shed light on our species' recent, frequently convoluted history and how it affects the genetic makeup of disease. The effect of our evolutionary past and its consequences for understanding human disease can no longer be ignored by medical practice; evolutionary approaches must inform medicine. This is especially true with the development of clinical whole-genome sequencing and personalized medicine.

Rise of personalized medicine

The practice of medicine has truly undergone a paradigm shift from the largely "reactive medicine" of the past to a more "proactive predictive medicine" focused on disease prevention. Instead of treating a disorder, there is a shift towards the individualized, data-driven care of each patient.

Instead, the goal is to approach each patient as a unique case and incorporate a range of tailored information, such as genomic, epigenetic, environmental, lifestyle, and medical history. The accumulation of these data into a customized virtual representation of the patient is intended to assist in the patient's rational therapy design by combining predictive modeling based on past experiences (**Figure 1**). The goal of precision medicine is to create computational models that incorporate clinical and basic scientific data and information in order to understand the disease from a mechanistic perspective and to support individualized therapy choices (3).

We now possess a vast arsenal of treatments for all major ailments, thanks to the 20th century. However, therapy frequently fails to be therapeutic and may also have serious negative effects. Furthermore, the widespread usage of these medications has exposed significant inter-individual variations in therapeutic response. In addition to age, sex, nutrition, and environmental exposure, there is evidence that a significant amount of the diversity in medication response is genetically determined. As a result, attention must be given to efficient treatments for smaller patient subpopulations who exhibit the same disease phenotype but have unique genetic profiles (7).

Next generation sequencing

Personalized medicine means using the appropriate drug at the appropriate dose and timing for the appropriate patient. Identifying the disease-related and drug-sensitive genes and SNPs at multiple levels, followed by extensive genetic screening to find those variants on each individual genome, is the first step towards achieving personalized medicine. NGS has been used to identify these variants in the human genome and epigenome, including those variations for illnesses, and has significantly advanced the development of personalized medicine (8). It enables researchers to better uncover the genotype-to-phenotype causal processes when combined with other experimental investigations (2).

Biomarkers for diagnosis, prognosis, and treatment are being investigated for many diseases through the study of essential NGS data and other related data. The associated new findings will greatly enhance patient care and enable drug development (9).

The enormous volume of NGS data significantly increases sequencing coverage; however, processing the enormous dataset efficiently requires greater CPU resources and more efficient bioinformatics tools (**Figure 2**). Additionally, short-read mapping and de novo assembly are made difficult by the short length of NGS reads. The abundance of repetitive and homologous sections seen in eukaryotic genomes, particularly those of mammals, can induce alignment problems and assembly collapse. The challenges of accurately conducting short-read alignment, de novo assembly, SNP calling, RNA-editing identification, gene fusion detection, and other NGS applications are further exacerbated by the sequencing errors of NGS. Along with advancements in linked bioinformatics techniques, NGS technologies are constantly evolving. In the future, several characteristics of NGS technologies, including the short read length, sequencing depth, substantially higher sequencing mistakes, sequencing cost, and sequencing time, will be significantly improved. These developments will make it much easier to use NGS technologies in the field of personalized medicine (2).

Translational bioinformatics

A potent technique for bridging the gaps between the systems biology research and personalized medicine is

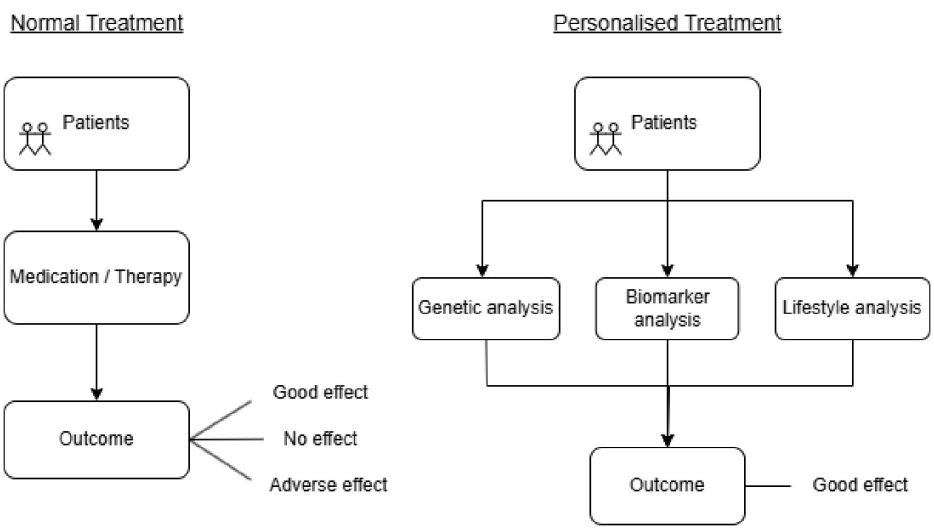

FIGURE 1 | Normal treatment vs. personalized treatment (5, 6).

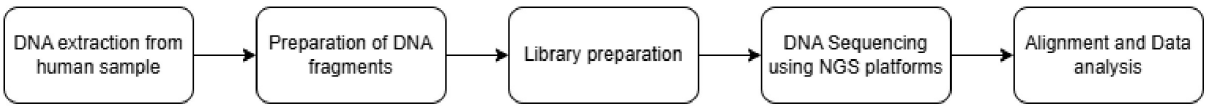

FIGURE 2 | NGS steps (10, 11).

translational bioinformatics. To solve issues and enhance the communication, understanding, analysis, and managing of biomedical information, bioinformatics uses computational methods. Translational bioinformatics, according to the American Medical Informatics Association (AMIA), is a new discipline that optimizes the transformation of increasingly substantial biomedical data, and genomic data in particular, into proactive, predictive, preventive, and participatory health. For instance, poor clinical workflow management, ineffective communication, and a lack of centralized data management are among the most challenging and important aspects of clinical outcome evaluation (9).

Applications of biomedical informatics in translational medicine would allow for efficient work flow management in both research as well as clinical environments. Clinical and laboratory data streams would be better integrated with the help of translational bioinformatics. Applications like concept representation and electronic record architecture would make it easier to communicate information and establish, implement, and adhere to the standards in 169 Translational Bioinformatics and Systems Biology Approaches for personalized medicine. Furthermore, translational bioinformatics can aid in the efficient use of healthcare resources and the reduction of clinical risks. For example, the most affordable and effective method of avoiding unfavorable treatment instances is computer-based information systems.

Translational bioinformatics can, most crucially, encourage the use of personalized medicine. It will enable researchers and medical professionals to create individualized plans for getting the appropriate medications in the right dosages to the right patients. Such strategies will enhance communication amongst multidisciplinary groups and assist in overcoming therapeutic resistance and adverse outcomes (7, 9).

Conclusion

The more accurate results produced by bioinformatics techniques enable reliable interpretations. The field of bioinformatics has perspectives that include contributions to the study of the human genome that result in the development of novel drugs and targeted therapies. To comprehend biological processes and the subsequent evolution of human well-being, it is crucial that bioinformatics and other disciplines work hand in hand. Integrative and translational genomics present new problems for bioinformatics today that will ultimately result in personalized medicine. The ongoing research in these fields aims to equip scientists with a much expanded set of computational tools that make it easier to convert the acquired data into biologically relevant knowledge. In a nutshell, translational bioinformatics is crucial in the development of personalized medicine from systems biology and pharmacogenomics. Translational bioinformatics can be used from the biological and informational perspectives to accomplish this goal.

On the biomedical side, translational bioinformatics would make it possible to identify biomarkers based on systemic evaluations.

On the informatics face, translational bioinformatics techniques built on data fusion, data mining, and knowledge representation can enhance clinical and academic decision-making.

Overall, we are moving into a new era of data-driven health care. The use of bioinformatics techniques continues to significantly improve patient care. Infrastructure, information technology, policy, and culture need to catch up with some of the technological breakthroughs. There are numerous opportunities for researchers engaged in cutting-edge translational bioinformatics, and the future appears promising (7).

Future potential

With the invention of molecular cloning techniques and the automation of DNA sequencing techniques, bioinformatics has advanced substantially. Large-scale sequencing of genomes and transcriptomes started with the development and use of new generation sequencing platforms. This contributed to the development of bioinformatics methodologies and tools at a level outside of academic centers, including forensic biotechnology, agricultural biotechnology, gene therapy, and animal and environmental biotechnology. Currently, genomics, proteomics, metabolomics, transcriptomics, and molecular phylogenomics are some of the fields where bioinformatics is most useful (12). The production of biomarkers will enable the development of safer and more individualized medications, which will further the advancement and use of bioinformatics. With the help of bioinformatics, efforts involving the sequencing of individual genomes and metagenomics will proliferate in the upcoming years. The future of bioinformatics, in our opinion, will involve

specialization in many fields that go deeper into science, down to the level of nanopores and even the atom itself (2).

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