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Chapter 12: The future of genomic medicine: A vision for equitable, data-driven, and artificial intelligence-augmented personalized healthcare

12.1 Introduction

Over the past two decades the cost of sequencing a human genome has dropped 99.99999995%, down from the $3 billion project completed in 2003, from costly $300 million projects in 2007 to just several hundred dollars in recent years. In 2013, a leading sequencing company introduced a new machine that costs just $1,000 to generate a whole human genome. With advancements in sequencing technology and analytics tools, as well as the rapidly increasing size of genomic data being generated, it has been predicted that genomic information will cover every citizen on this planet. The growing expectation is that, with the widespread utilization of genomics, more data-driven and precise healthcare will be achieved, diagnosing diseases earlier, suggesting better treatments, and preventing diseases before they occur.

A vision is posited for imagining the future of continued advancements in genomics from an analytical perspective. This vision is for a future where genomics becomes a key driver of healthcare and introduces a more equitable, data-driven, and AI-augmented healthcare system. Projects and works in realizing this future are presented, from academic research analysing large-scale sequencing data, including deeply understanding the genetic basis of common and rare diseases and the development of more accurate health risk prediction models, to interdisciplinary collaboration with businesses on utilizing genomics to evolve and evaluate new diagnostics and treatments. There will be a new section on the established genomics research facility, engaging and collaborating with the large healthcare and health technology sector in applying genomics to improve and evaluate current health services. At the end of the essay, the discussion will be shifted towards presenting and reflecting on the resulting works, including both significant improvements and important challenges in current practices, and issues needing extra attention.

As new opportunities emerge with genomic technologies, a critical consideration for healthcare is their distribution and accessibility. There is an ongoing debate in the US about whether genomic medicine will follow a similar course as family planning advancement, leading to disparities in successful intervention rates among diverse populations. In the context of genomics, diversity often refers not only to the wide range of genetic makeups but also to social, economic, and other factors that are intertwined with genetic backgrounds. In the landscape of health genomics, from testing and interpretation of genetic data to the development and application of treatments informed by genomics, there is a growing concern that the benefits of genomic medicine are expanding more dramatically to some subpopulations while leaving many others behind, resulting in prolonged disparities in diagnosis and treatment for a wide range of diseases.

Fig 12.1: Data-Driven, and AI-Augmented Personalized Healthcare

12.2. Understanding Genomic Medicine

In the landscape of modern healthcare, genomic medicine is often discussed in terms of its transformative potential, from enabling the provision of patient and disease "targeted" care, to fundamentally shifting the ownership and economy of medical knowledge. However, the relevance and breadth of such a vision can often feel difficult to fully comprehend, for both patient and provider alike. Before examining genomic medicine in specific regards to data's role and AI's application, it is useful to firstly provide a thorough overview, to establish a framework with which to understand the complexity and potential of genomic medicine as a whole, and its subsequent realization.

Genomic medicine is the application of an individual's genetic information to predict susceptibility to diseases and response to treatments, and the subsequent use of this information in the diagnosis and treatment of diseases. Diseases arise through the complex and often interacting genetic architecture of an individual, and so the elucidation of each individual's genetic makeup opens opportunities to more targeted diagnosis and treatment of disease. Understanding of the importance of genetic information to health is long standing; a 1957 article warned of "the rebirth of eugenics" that would be ushered in by the discovery of the DNA. In recent years there have been substantial technological and research based advancements in the capability of reading and understanding of the human genome. However, as shown through a tool which compared patient recorded symptoms to a database of health records to provide potential diagnoses, even a most recent understanding of the genome remains only a base level upon which to build effective treatments.

12.2.1. Definition and Scope

This subsection will address various societal, technical, and ethical challenges that must be met for the equitable, data-driven, and AI-augmented future of genomic medicine to be realized. It will also touch on pivotal research areas that must be expanded. Addressing these issues will be crucial for ensuring that the coming transformation benefits every patient who can be helped by it. The scope of this text is primarily focused on the clinical application of germline genetic variation.

A concurrent initiative proposed a broader definition of genomics relative to the focus on genetic and molecular research. Since then, in both practical and academic contexts, 'genomic medicine' has commonly been understood to encompass the use of genetic information for preventive, diagnostic, and therapeutic approaches to medical care that are tailored to individual characteristics. A focus on precision or personalized medicine has also become increasingly widespread outside of genomics, envisioning a shift from traditional one-size-fits-all medical treatments to a more targeted and tailored therapeutic approach. Genomic medicine thus understood is a highly multidisciplinary field that draws together genetic science, health informatics, and public health to improve patient outcomes through more individually tailored healthcare based on genetic information and predictions of population and individual disease risks. There have been substantial advances in knowledge and capacities to deliver personal genomics services, particularly direct-to-customer testing for ancestry tracing, pharmacogenomic response profiles, metabolic traits, and various health and reproductive risk tests. Nonetheless, the application of genomic knowledge to routine primary care remains particularly challenging to deliver, the complexity of which is exaggerated by primary care being understaffed, under-resourced, and generally slow at adopting new technologies. This text will define the scope of those diseases referred to here as being inherited or hereditary, before outlining six broader health sectors in which genomic medicine is expected to have transformational outcomes. The targeted development of at least the following six medical specialties should start (Challa, 2022; Chava, 2022; Chakilam & Rani, 2024).

12.2.2. Historical Context

As ancient as Hippocrates' famous quote, "It's far more important to know what person the disease has than what disease the person has" shared around 380 B.C., the idea of personalized healthcare has always been a deep-rooted yet idealistic one. Although significant advances in genomic understanding have been made in the past decades, it is still extremely challenging to deliver the right treatment at the right time to the right person. As a medical discipline, genomic medicine that relies on genomic information to stratify disease risks and individualized patient care has recently gained unprecedented traction. Understanding historical context is particularly essential when studying the prospects of emerging fields such as genomic medicine. With a timeline built upon pivotal findings and key challenges faced by researchers and practitioners over time, this innovation aims to provide a comprehensive look into the ongoing evolution in genomic medicine. This timeline will be enriched by downstream analyses exploring different ways that lessons from historical successes and failures can inform current and future approaches. Whether genomic medicine is still a field in its infancy is debatable, yet its trend as a new frontier of medicine oriented by rapidly evolving technologies is undeniable. Genomic medicine has never been as important as a facilitator for delivering healthcare during the prolonged course of the HGP. The information and resources generated have revolutionized, and will continue to fundamentally reshape biomedicine, medical practice, and public health. It is a good time to envision the future direction for genomic medicine as the completion of the first reference human genome is expected. Major points of consensus among the five discussants were summarized, and six prospective articles drafted as the product is still futuristic vision and embassy. The conformity between these articles and the current situation, of course, may vary with the inevitable development of genomic medicine. However, the current critical review seems in retrospect to be more an inquiry of what genomic medicine should have been then, and what it should be now.

12.2.3. Current State of Genomic Medicine

This subsection presents an assessment of the current state and future prospects of genomic medicine. After a brief overview of the field, the opportunities for interdisciplinary research and clinical care recommend a greater engagement between bioinformatics and clinical specialties. Advances in genomics and related fields of '-omics' research have facilitated great strides in health and medical research, and have opened up new possibilities for personalized healthcare. Ultra-high-throughput technologies have emerged for the experimental investigation of biological regions, molecules and their structures, and computational methods have similarly grown apace to analyze the data arising from these investigations. Prominent among these have been cDNA microarrays and more recently next- as well as third-generation sequencing technologies, together with associated bioinformatics methods. As a result of the precipitous cost reductions for sequence determinations since the completion of the Human Genome Project, these technologies have also had a profound impact on clinical and medical research and practice. This is of great interest not only for human health, but also for agriculture and biotechnology, and has led multiple governments and companies worldwide to fund large-scale genome projects to investigate the biological basis of health as well as disease susceptibility. A major aim of these projects is to foster the development of 'personalized' medicine, that is, to provide the most effective therapy with minimal side effects tailored to an individual's molecular and genetic profile. Various initiatives are currently underway exploring the use of genomic, transcriptomic, epigenomic and proteomic information to personalize different fields of medicine, to predict the patient's response to a certain treatment, to detect contagious diseases, or to screen for infant genetic diseases, or certain genetic backgrounds which are linked to disease susceptibility. In this context, pharmacogenomic studies investigate the genetic variability of drug metabolism, and especially its effect on the efficiency of the pharmaceutical substance, its toxicity, tolerance, and adverse or allergic reactions. These studies aim to predict an individual's response to a drug before medication, and to optimize the dosage or change the drug for a more effective and less toxic therapy. While genetic tests are currently administered only as a last resort in case of diseases with known genetic causes or for the detection of transmissible diseases among progenitor couples, the spread of appropriate technology and the significant reduction of costs respectively is accelerating the application of genetic profiling to the most disparate medical issues.

12.3. The Role of Data in Genomic Medicine

Genomic medicine revolutionizes healthcare by leveraging comprehensive datasets to personalize treatment based on individual genetic make-up. It has wide-ranging applications from repurposing drugs to designing cost-effective, cellular immunotherapies against cancer. Understanding the differences in data types, challenges, and goals is essential for developing a successful model or personalized treatment (Chava, 2022; Komaragiri & Edward, 2022).

In healthcare, decisions rely on the understanding of various data types. Genomic models use data collected from genome-wide association studies, whole genome or exome sequencing, and gene expression. There is also data concerning a patient's clinical environment such as family history or available demographics. Different models need different data.

The quality of the data dictates the quality of the model or the treatment. Missing data affect model performance. There may be a model seeking to predict something that happens much later in time and hence normal monitoring is impractical. In that case, data may need to be collected as often and as comprehensively as possible.

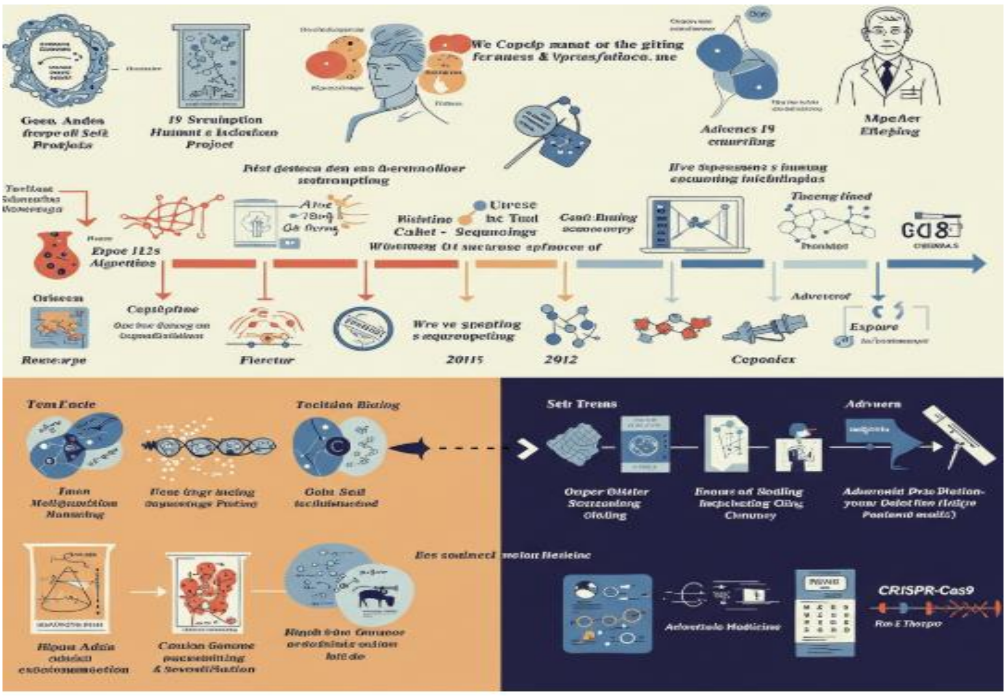

Fig 12.2: Evolution of Genomic Medicine

12.3.1. Types of Data Utilized

Genomic medicine has the potential to deliver personalized approaches to diagnosis, prevention and treatment. Different types of data are essential for understanding health outcomes. Genomic data can be used to predict and treat disease based on knowledge of an individual’s DNA. Clinical data provide information on patient medical history, family history and recorded phenotypes, and can reveal patterns that forecast a potential health issue. Combined with other molecular-level data types, environmental/lifestyle data can provide a more comprehensive understanding of health outcomes. However, such data are often treated in isolation, and the benefit of an integrated approach is limited. By categorizing data types and outlining their roles and contributions, the reader will be more aware of the various facets required to advance the field. The synergy between data types is illustrated through the application of integrated data, highlighting how the analysis of combined data can deliver an enriched model of personalized healthcare. The discussion will also touch on challenges, in particular how data heterogeneity complicates integration and limits the transfer of knowledge between different data sources and study designs.

Genomic medicine stands on the edge of revolutionizing personalized healthcare through early diagnosis, prevention, targeted treatments, and monitoring patient health outcomes. The understanding of health outcomes associated with genomics eases the interpretation of genomic data and enables a predictive perspective on health, i.e. the possible analysis of the risk of future states, such as disease. However, while genomics is a powerful lens through which to interrogate health outcomes, different types of data are essential to give a complete picture. To date, multiple applications have separately used genomic or clinical data in the inference of health outcomes but to a large extent treat these data in isolation. The benefit of an integrated approach is evident from the range of applications linked, which is diverse and demonstrates a broader scope than when separate data types are studied. Furthermore, the implications of health outcomes on specific data types tend to have a broader scope when multiple facets are considered, for example, building discussion around the treatment of asthma in the context of genome-environmental interaction, and expanding a conversation from purely genomics. Subsequently, this signals a greater potential for transdisciplinary work in epidemiology and the development of guidelines based on integrated data.

12.3.2. Data Collection Methods

This subsection elaborates on data gathering in genomic medicine, its significance for quality and reliability, as well as addressing the challenges. High quality and reliable data are central to genomic medicine and its advancement. In the context of clinical associations, the quality of genetic data is equally important as the quality of clinical phenotypes. Using advanced technologies, genetic data are generally more accurate; however, possible imprecision needs to be considered. As for data collection methods used in clinical or study contexts, the reliability of data can be regarded as being more directly controlled than its quality.

Advanced technologies in genomic sequencing and bioinformatics ensure an accurate identification of genetic data, covering both the typed SNPs and the imputed dosages. There are plenty of ethical considerations in the processes of data collection, storage, and analysis. It is important to evaluate what kind of ethical requirements need to be fulfilled before collecting the data, making them public and/or associating them with other datasets. The respect of patient informed consent is one of the main requirements for these purposes. Several methodologies can be employed for the collection of genetic and clinical data in the clinic and research environment. In clinical contexts, mainly individual genetic testing is conducted, providing the patient's genetic biomarkers only, and support is offered solely to the patient.

Resistance might be met from healthcare providers, especially if prescribing off-label drugs based on N-of-1 predictions, or from patients, if they perceive their standards of care fall below those received by the overall population. The adoption of personal genomic services in public health context might also be hindered because of representativeness in the datasets harnessed.

12.3.3. Data Privacy and Security Concerns

Given the sensitive nature of genomic data, there are ongoing efforts to create and enhance legal and technical frameworks to ensure robust privacy and security, while it is reasonable to assume that it has a unique nature and potential vulnerabilities to threats. Moreover, there are various jurisdictions globally that apply distinct legal frameworks on genomic data, each of which attributes specific features differently. It is important to address the critical issues of privacy and security within the landscape of genomic medicine, as well as on a global scale. Attention must be paid to various legal and ethical perspectives and consider the socio-technical challenges to complement the current debate and examine the trade-offs and, likely, forthcoming developments. Even shared between healthcare providers, patients, and researchers, improves the detection and treatment of diseases. However, there are also ethical challenges, as healthcare practitioners must balance the potential benefits of sharing data with respect to public health and scientific advancements with the privacy protection that individuals expect. Moreover, the sharing of genomic data can also lead to unintended harms. For instance, the discrimination of individuals based on their genetic information. There is evidence that insurance companies may misuse genetic information to deny services, or otherwise discriminate against individuals. Indeed, there are cases wherein companies hold and refuse to share genetic information in the fear of its misuse. As genomic data becomes more prevalent and useful, there is a need to address these ethical issues and introduce measures that prevent misuse.

12.4. Artificial Intelligence in Genomic Medicine

Artificial intelligence (AI) has the potential to transform genomic medicine, a field that generates extensive genomic datasets. Such datasets often include imaging, genomics, epigenetics, and transcriptomics. AI algorithms have been developed that can interrogate such large datasets using a variety of data types and modalities and construct complex models. These AI-based models can uncover new insights from the data, make predictions, and suggest further experiments. In genomic medicine, these predictions may lead to beneficial clinical actions. For example, AI has already been successfully used in variant interpretation by constructing complex discriminative models with a variety of different data types. This includes recent approaches that convert multiple data types to image format so they can be analyzed by cutting-edge convolutional neural network architectures. AI models of breast cancer pathways have been constructed that consider protein interaction networks, gene co-expression, and gene knockout data. The models predict the impact of novel coding variants with 99.3% accuracy reported as a receiver operating characteristic area under the curve. The AI models have been used to interpret pathogenicity of coding variants present in the ClinVar database that was uninterpretable by the current best variant effect predictors and identified three potentially novel variants in three likely breast cancer susceptibility genes. AI has also been successfully applied to predictive modeling in genomic medicine.

However, efforts driven by the application of machine learning technologies tend to focus on algorithmic considerations, while less attention is paid to the nature of underlying features and to important quality assurance and control challenges. A computational systems biology approach is illustrated that augments standard tools and resources used for genomic and genetic analysis with artificially intelligent models mechanistically grounded in current understanding of pathobiology. The approach is applied to the automated analysis of head and neck cancer next-generation sequencing data. Since the results emphasize the drivers of aberrations that can spur development of targeted and immuno-therapies, they demonstrate the great potential of artificial intelligence to support progress in precision medicine. However, it is also noted that ongoing efforts and collaborations are needed to address emerging challenges: the issues of data bias and the lack of algorithm transparency. Only a comprehensive effort comprising broad expertise in biosciences, bioinformatics, engineering, and artificial intelligence could deliver effectively and responsibly upon the promise of artificial intelligence technologies for contemporary biomedicine and healthcare.

12.4.1. AI Algorithms and Their Applications

Artificial intelligence (AI) algorithms in genomic medicine have generated a broad spectrum of applications. This includes identifying genetic variants associated with clinically meaningful phenotypes and in silico drug screening to guide treatment plans. Machine learning models have been developed to predict health outcomes based on genomic data. In clinical settings, they can be used to stratify patients for targeted interventions or to analyze probabilities of health outcomes of interest in real time. Machine learning boosted Cox models have been implemented and correlated single nucleotide polymorphisms (SNPs) that were significantly associated with clinical outcomes of interest were interrogated. Analysis of generated SNPs demonstrated high agreement with published results of known associations. An application that allows the querying of multiple genetic variants to concurrently analyze their association with a set of phenotypes of interest was showcased. Lastly, an application that helps the ethambutol dose of tuberculosis (TB) patients with a high risk of visual impairment was presented, allowing to optimize an otherwise complex and lengthy treatment plan. Additionally, the potential of Natural Language Processing (NLP) to support the processing of large volumes of unstructured routine clinical data, including free-text patient medical history or clinical notes, to enhance genomic data interpretation was highlighted. Moreover, it was demonstrated how the implementation of NLP can substantially streamline the discovery of clinically actionable genetic variants signatures derived from complex genomic data on a broad patient population. To facilitate the discussion, example applications that pertain to genomic data are showcased, though other datasets and purposes are equally valid. It is encouraged that all output tables or figures be validated. Regardless of the algorithm type, the accuracy and completeness of the results output are paramount to ensure meaningful and reliable associations are identified and avoid overinterpretation of spurious or incomplete predictions. In this light, best practices for algorithm validation and potential avenues for output verification are discussed. Additionally, specific pragmatic recommendations for implementing ML models are offered, including sampling, data formatting, and data preprocessing. Finally, recurrent challenges potentially hindering wider and more efficient integration of ML models into routine clinical workflows are highlighted. Recognizing the importance of interdisciplinary efforts across medical, computational, and engineering domains, collaboration with various investigators and project teams with the aim of discussing open problems and suggesting areas for continuous improvement and innovation widespread applicability.

12.4.2. Machine Learning in Genomic Analysis

The intersection of machine learning and genomics could delineate the role of the genome in health and disease. Machine learning, specifically deep learning, can be vital in understanding genomic sequences. Machine learning comprises a section of artificial intelligence where algorithms learn patterns and derive insights from data. Since genomic data is high-dimensional and complex, machine learning methods are ideal.

They surpass conventional statistical models that can solely recognize straightforward linear relationships. Recent studies have utilized machine learning to assess changes in DNA methylation and gene expression more comprehensively regarding complex disease phenotypes. In cases where non-linear relationships exist between these omic modalities, more sophisticated machine learning methods like neural networks and random forests can demonstrate novel insights. Broadly used machine learning in omics is the supervision of models for anticipation or stratification. One space with massive promise for machine learning in genomic research is large-scale biobank programs. Machine learning is expected to be transformative in all realms of biobanking, but more so in genomic medicine due to the comprehensive, high-quality, and multimodal data being assembled in these endeavors. Analyzing this data with machine learning can unearth intricate and multi-tissue transcriptomic architecture or improve the current understanding of the genotype-phenotype-environmental (GixEn) interactions driving a specific phenotype. The genetic influence is not isolated to a specific feature but is shared broadly across traits. An examination of the data has density plots reflecting the genetic effect sizes for nine traits. Shared genetic influence is noted for blood traits, mental health, and autoimmune diseases like type 1 diabetes, supporting known genetic and pathogenic mechanisms between some of these diseases. Colocalization analysis supplements these results, indicating that many reported trait-associations on certain phenotypes share genetic origins with gout and other traits as well. Unequivocally identifying these shared mechanisms is difficult in a hypothesis-driven framework but may be possible if machine learning were applied. However, the accuracy and performance of these models are directly relevant to the availability of adequately large training sets, a challenging requirement for omics modeling in general, particularly for mechanistic or causal inquiries. Furthermore, the availability of commercially accessible -omics databases is sparse and machine learning reliable. There is a vital call for novel machine learning approaches and collaborations between machine learning and biology in research and panel discussions. This call is supported by a brief review of the opportunities in basic, clinical, and bioethics research. Machine learning methods are used to derive insights from complex data regarding complex disease phenotypes. The comprehensive assessment of changes in gene expression, DNA methylation, and PiH in bipolar disorder demonstrates how these traits relate in control subjects and patients diagnosed with a mental health condition. Despite intra-modality non-linear relationships, the relationship across all modalities is largely linear. However, given the high-dimensional and complex nature of these relationships, conventional linear QTL methods are not able to resolve important PXI or GXI interactions. Machine learning methods offer a culturally important view of the anatomy of eQTMiH interactions or reveal new insights about genes, miRNA and (m)RNAs that modulate gene expression or modify the epigenomic or transcriptomic landscape of gene expression in a cell type. Novel biological findings obtained using advanced machine learning underscore the importance of this work. A machine learning method is developed to generate well-suited gene sets to test for differential expression or differential splicing. When these QTL are |5|These tests are conducted on well-known LD hub genes. Using a filter for G2P diseases, the average number of significant associations is tallied. This filter is permuted 108 times, and empirical p-values are plotted. When comparing this with the scheme of a network smoothing approach, differential expression associations are statistically significant, indicating that this proxy method may only work on this false scenario. Regardless of the method, it is observed that the differential expression approach reconciles significantly more results than differential splicing. On average, the novel prediction was well within a -/+ 1Mb window. Moreover, using Average ChromHMM States, a sizable number of predictions fell within active regulatory domains. To further develop this observation, according to the GTEx guideline, these two novel regulatory predictions should not overlap perfectly with gene start sites. From this standpoint, a demonstration allows confidence in the developed method to predict QTLs up- and down-stream of genes.

12.4.3. Challenges and Limitations of AI

The Future of Genomic Medicine: A Vision for Equitable, Data-Driven, and AI-Augmented Personalized Healthcare is presented. Since the completion of the Human Genome Project, the vision of integrating genomic data into routine clinical care, termed genomic medicine, has tantalized practitioners, researchers, and governments globally. This vision has met several milestones, including cost-effectively sequencing human genomes and multiple national and large institutional efforts designed to systematically build the evidence-base for implementing genomic medicine. Despite these successes, substantial work remains to realize the full potential of genomic medicine. Critical barriers to global implementation are presented alongside a vision for genomic medicine firmly embedded into health systems across the healthcare continuum. The goal of this article is to critically examine the current state of genomic medicine and the future direction of the field, with an emphasis on challenges to global implementation and a vision for addressing these hurdles. Since the groundwork for the Human Genome Project was laid in the 1980s, the vision of integrating genomic data into routine medical care, hereon termed genomic medicine, has tantalized practitioners, researchers, and governments globally.

12.5. Equity in Genomic Healthcare

While efforts to integrate genomic technologies into health care continue to accelerate, ensuring that the benefits are accessible to all is more essential than ever. This issue of the Future of Genomic Medicine series explores equity in genomic healthcare and the need for equal access across diverse populations. A vision is outlined for a more equitable healthcare system that leverages genomics to drive tailored, data-driven, and learning solutions for the communities that need them most. In collaboration with communities, actionable strategies and policy recommendations are provided for building inclusive genomic healthcare systems.

Genomic medicine raises questions of equity at multiple levels, from broad policy considerations for public health initiatives down to decisions taken by individual clinicians. It is important for a healthcare system to ensure that access to tests, treatments and services is equitable—that people who need or could benefit from these services access them in a timely way. There is established precedent, however, for health technology assessments recognizing that the use of a new technology has the potential to lead to inequitable access, consider the object or the provider of care, and that entire technologies may not be adopted if their use is anticipated to do so. There is a preference in a healthcare context to discuss equity, rather than the more abstract concept of 'equality'. This is consistent with an understanding of healthcare as a public service, and the state as having duties to provide this service..

Fig : AI in Healthcare

12.5.1. Access to Genomic Services

The rapidly decreasing cost of DNA sequencing, along with increasing research and public investment in the field of genomics, has democratised genetic information in a speed, scale and depth that far exceeds Moore's Law. As such, the general public's exposure to genomics has expanded to a degree that exceeds what most clinicians and public health practitioners are comfortable with. The field of human genomics has seen a rapidly expanding expert knowledge base, while it is simultaneously expected of the lay public to not only keep up with it all, but to also make sense of it for clinical decision making and well-being. This has generated a potentially hazardous knowledge delta. The rapidly expanding expert knowledge base in the field of human genomics has further spurred on the exponential growth in the field of health-related clinical genetics, adding new levels of complexity and stress to the clinical and public health disciplines that are expected to utilise the same expert knowledge base to guide clinical practice or derive public health benefit.

This exponential augmentation in knowledge would have been more manageable if it was not further complicated by the rapidly escalating multitude of genetic testing services offered via the DTC model. Even if the previous knowledge delta between the general public and the expert domain was never to be bridged, the burgeoning DTC industry is already expanding the knowledge gap between the genomics experts and other healthcare and public health practitioners in a way that no vocational development can reasonably remedy. With the rise of genomics in personal health management, it is crucial that there is widespread equitable access to all people, regardless of socioeconomic circumstances, across a range of healthcare settings. Ensuring access to genomic technologies, however, has potential to impact on health equity, a challenge that needs to be urgently addressed. There is an ongoing need to reduce disparities in access to care, particularly in the genomics era. This calls for methodological expansion in the design of health technology assessments with greater consideration to ethical, legal, and social issues to better evaluate the potential for health care disparities and inequitable access from individual genetic information.

12.5.2. Disparities in Genomic Research

A rapidly expanding body of research now underscores the profound disparities in health and access to healthcare that exist in the United States and globally. Studies have demonstrated that African Americans are twice as likely as whites to be diagnosed with diabetes and suffer from hypertension. Data also indicates that American Indian, or Alaska Native, adults are 50% more likely to have heart disease than white adults. These health disparities are multifaceted, with roots in the availability of healthcare, a population's overall health literacy, differing levels of exposure to environmental risks, and more. Furthermore, without a nuanced and multidimensional understanding of an individual's health, healthcare decisions themselves, which have life or death consequences, are not entirely informed. As a result of health disparities, there exist preventable differences in health outcomes, risk factors, or treatment effectiveness that disproportionately affect racial and ethnic minorities. As a result, there is a growing interest in addressing the causes of health disparities among researchers from diverse fields, including biology, sociology, and public health. Since the completion of the Human Genome Project in the early 2000s, an increasingly central role in efforts to ameliorate health disparities has been thought to be played by genomics. However, past and current genomic research related to health disparities is varied, prompting some to see it as a nascent field in need of further definition and development.

12.5.3. Strategies for Promoting Equity

As genomics becomes integral to patient care, it is vital to ensure that all populations have equitable and ethical access to this powerful information. Implementing four policy frameworks can advance equity in both research and health services. First, increase funding and resource sharing for research and genomic services in relation to underserved populations. Second, prioritize community and public education on genetic and genomic medicine through brochures, fact sheets, social media resources, and other print material on genetics and genomic services. Third, the healthcare system should form collaborative programs with link workers/lay health workers/community health workers and community organizations, including faith leaders. These community partnership-based programs provide essential education, support, and navigation tailored to the individual and enhance learning. Fourth, healthcare providers must be competent in cultural competence, maintain an awareness of patients' diverse backgrounds, attitudes, beliefs, and practices, and adapt communication to consider these factors. Encouraging providers to learn how to communicate respectfully and effectively using interpreters when necessary is vital. Healthcare providers should discuss clinical options emphasizing flexibility and address all patient concerns, hesitations, or uncertainties. Additionally, transparency of research practices and the value of consenting communities for understanding their role in the research process are essential to empowering individuals as well as community organizations to advocate for their best interests. Mitigating disparities in genomics services requires a systemic response. It is one of the many hurdles to providing such a response. With the emergence of the Precision Medicine Initiative, better and more systems are working on personalized medicine issues and there is a need for genetic research. The Guide to Community Health Improvement provides a starting point with information on health priorities and root causes. Although the Guide helps, it fails to provide information on quantitative differences or risk factors over time. Furthermore, guidance is needed on how to understand and use the vast amount of already available data to justify actions and drive change.

12.6. Personalized Healthcare Approaches

In the future of medicine, wearables will enable the passive collection of vital signs, heart rate, and activity levels. Combined with lifestyle information and current and historical medical records, these devices will provide rich insight into a wide range of aspects of human health, potentially allowing for the early detection of a range of health conditions, from metabolic diseases like diabetes to neurological disorders such as Alzheimer's, even in asymptomatic individuals. With ever larger and more diverse sets of data, new machine learning models will be able to generate precise and personalized health advice, enabling the development of preventative treatments that are tailored to each individual in terms of both the risk profile and the genetic background.

For those already suffering from disease, the advent of genomic medicine and personalized healthcare can significantly impact patient outcomes. The identification of the genetic factors associated with an individual's disease can enable the development of effective treatments that are tailored to these genetic profiles. This has direct consequences on the choice of medications that might be most effective in that setting. However, perhaps more importantly, this switch enables a more centralized DVDC design. This, in turn, can help transition the current model of care, which can be characterized as prescriptive, to more patient-centric educational models. The participation of the patient is an essential missing link in the current setting – a doctor can provide a list of possible treatments, but if the patient does not understand the implication of each choice the chances that the correct course of action will be followed decrease dramatically. It is therefore of utmost importance to foster collaborations in initiatives that aim the education of the patients, so that they are in a better position to understand and follow complex treatment plans. This need is even more pressing in the case of dialysis treatments, which are burdensome and have substantial impact on a patient's daily life, as compliance to the proposed course of action is a crucial factor that is strongly related to the outcome of the treatment.

Examples of personalized treatment protocols include the selection of medication and dosages for patients with cancer based on the genetic profile of both the patient and the cancer, the use of gene-altering drugs as responses to particular genetic mutations and creation of 3D-printed drug-filled capsules, or the direct injection of a drug candidate into an eyeball, a protein that would not be functional or well understood without having structural information. There is substantial potential associated with personalized healthcare, but also significant problems that must be addressed to reach this potential. Most notable of these is the high cost of many treatments, indicating that some may not be feasible to be made available to the general population. Nonetheless, this and many other challenges (e.g., issues of privacy, sustainable data security, the need for substantial better understanding of biology, and many technical issues related to the manipulation of genetic material) can be addressed via collaborations between pharmaceutical companies, regulatory agencies, and academia. These should aim to foster and accelerate the development of these and other emerging technologies and their corresponding treatments .

12.6.1. Tailoring Treatments Based on Genomic Data

As genomic data become less expensive and more widely available, the ability to tailor treatments based on these data holds vast potential for improvements in health outcomes. Human biologies differ in profound ways, due to genetic variations and other factors, including but not limited to immune responses, dietary choices, metabolisms, and gut bacteria. As these factors influence the responses of individuals to treatments, by analyzing the genetic profile of a person it is possible to design a treatment plan maximally efficacious for that individual. This includes choosing the most suitable drugs at the most appropriate dosages, as well as suggesting those treatments with the best cost-effectiveness profile. Approaches of this kind can be considered as a form of applied pharmacogenomics. Though study in this matter is not new, interest in the field has increased dramatically, considering the unprecedented possibilities provided by emerging data and machine learning technologies.

For example, in the realm of chronic obstructive pulmonary disease, traditional symptoms-based treatments can fail to alleviate ill conditions in patients. While there is no cure, genome study promises the possibility of precise interventions, potentially preventing a serious lung disease. Investigations in pharmacogenomics publicize several cases of personalized treatments which were efficacious, while generic treatments had failed. Given all these contexts, it seems likely that the tailoring of treatments, or personalized healthcare itself offers vast potentials for public health. The sharing of data between healthcare services and genetic testing companies, the development of machine learning models, and the use of these models alongside existing electronic patient records, and the potential implications of such developments for current medical conditions and patient care on a broader scale are also discussed. However, several challenges need to be addressed in order to bring this potential to further fruition, including a better understanding of how best to integrate genomic data into practice, and thorough engagement with important ethical concerns.

12.6.2. Patient-Centric Models

The field of genomic/precision medicine holds the promise of more personalized and effective care. Although equity has been a key concern since the onset of the Human Genome Project, this promise is threatened by a wide array of issues surrounding unrealized equitable impact that span research and clinical implementation, from access to services to informational health inequalities. Efforts to render these advances accessible, equitable, and effective can be aided by a future-focused dialogue that includes diverse stakeholders.

Health inequities are pervasive and persistent. Today's most advanced genomic findings emerge from populations generally well-represented in scientific research, with an estimated 78% of genomics-based publications originating in studies of individuals with European ancestry. Clinically-available genetic tests similarly have much greater representation of and utility for European-descent groups; a review of genetic testing technology marketed in 40 European countries found that services were not available or not performed on site in the capital city of 57.5% of the countries. Without intervention, many current forms of ill representation are likely to persist. Major technological, conceptual, and infrastructural changes are required simply to improve the study of non-European ancestries let alone appropriately diversify findings. In any case, the nature of genomic insights means that there will always exist results which simply are not generalizable across populations. More fundamentally, a focus on population-level generalizability is somewhat inappropriate for a technology that is at its core about personal or individual level information.

12.7. Future Directions and Conclusion

Robust research on genomics has transformed the biomedical landscape and reinvigorated the clinical examination. This upturn has been particularly conspicuous over the last decade. With developing technologies, the base combined understanding of chromosomes, RNA, DNA, and proteins has percolated into a deeper understanding of biology. As a result, the capability of data manufacture and shift of empirical investigations has expanded significantly. These advances have spawned significant improvements in medicine, including the identification of novel drug targets, the improvement of diagnostic methods, and the propelling of personalized treatments. As clinical genomics and genetic services are progressively integrated into healthcare, it is fervently hoped that such advances will broaden and that all the populace will gain from those renewals. Nonetheless, contemporary diagnoses and treatments based on genomics disproportionately profit certain groups while discriminating against others. There are more than a few factors intensifying and reproducing this effect. This problem requires adroit handling via the ideological imperatives of an integrative and community-based discipline that promotes co-innovation and co-ownership between disciplines, community-based institutions, and populations at the grassroots level. Unconventional stratagems of multidisciplinary conferencing are indispensable for unloosing such co-innovation and co-ownership.

Health is determined by manifold biological, genetic, environmental, and social factors. Because genomics studies the DNA at the foundation of those factors, information compiled from the sequence analysis of the whole genetic material can uncover a wide range of genomic traits. Some of these traits have a direct connection with health, like inheriting disease risks or modeling unfavorable drug responses. Others have a connection either with health or relevant with lifestyle, and environment, like the likelihood of getting depressed or preferences regarding certain diets. As an overall view it can be depicted that genomics, and especially whole genome sequencing, has either a direct or an indirect impact on most clinical or preventive medical disciplines. This wide scope, together with the simplicity of data storage and processing, allows the establishment of a personal database maintaining health and personal data from all possible disciplines, and provides digital storage of EHRs. With the recent advancements in computational efficiency and utilizing AI applications, the integration of primary physicians, a wide range of specialist physicians as well as public health analytical staff to make better use of this database with mechanistic and individualized preventive health models will be feasible.

12.7.1. Emerging Technologies

To appreciate the future of genomic medicine is to appreciate the technology that will shape it. There is perhaps no more apt time to envision emerging tech that could lead to eradicating the prevalence of molecular biological insight than now. With global advances on the cusp of transformative potential, the field of utile biotechnology is evolving more rapidly than ever imagined. The arc from the PCR revolution to the lamentation of the cost of the first human genome was a testament to the capacity for change but a mere glimmer of the heady days ahead. Similarly, venture biotech investments still quaintly peaked in ambitions first reaching to the billion-dollar mark but now are hedging bets in expectation of multi trillion-dollar industries.

A subsection here is dedicated to presenting a superficial nod to appraisal of the technology evolving in concert with genomic informatics that appears the most promising factors poised to shape the future of genomic medicine. This selection is not exhaustive, but due to the puerile nature of this domain of biological exploration, the relationship between the tech and the informatics is concentrated. And although the narrative emphasis is on the prospects future tech offerings can provide and frame within the capacity of bioinformatic advancement in genomic medicine, it is important to remember: there are social and political realities, ethical conundrums, and cautionary warnings to consider as well.

The selection focuses on gene editing technologies; the most prominent to date are CRISPR/Cas9, but the window allows historiography through early RNAi manipulations, and consideration now of alternative CRISPR systems, Anti-CRISPR systems, and strategies such as prime editing. It is anticipated editing technologies will drive the eradication of many diseases with a genetic etiology such as thalassemia, cystic fibrosis, and muscular dystrophy. Although most curative applications at first will unlikely see genetic modifications within the germline, there is promise for future generations through more complicated manipulations. With the fifteen-generations per day mental backlog calculation, CRISPR now holds all 64 bit cassette recognition; this offers the possibility to tailor the bacteriophage defense systems or the transcriptional regulation components of gene engineering with absoluteness never before imagined or envisaged.

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