Personalized Health Care Decisions Powered By Big Data And Generative Artificial Intelligence In Genomic Diagnostics

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Abstract
Genomic diagnostics provide an essential tool for clinical decision-making since diseases can occur due to alterations at specific locations in the genome, especially when uncommon in prevalence. Genomic data are inherently complex and large, increasing the general need for sophisticated decision-support systems. Advancements in the further digitization of data and genomes, combined with efforts for closing the data collection gap, are generating enormous multidimensional datasets in this area. In general, potentially if volunteered by patients, the majority of the data is health-related. Novel and neglected but rapidly evolving technologies, including generative artificial intelligence, are currently enabling unprecedented opportunities in terms of automating complex and lengthy explorative data analyses. Actionable, health-related insights, which can be generated and interpreted by patients with increasing confidence from cherished or trusted digital hobbies outside the medical field, have the potential to more realistically change health behaviors. The ever-increasing data availability, as well as the increasing amounts of metabolomics, proteomics, epigenomics, and other 'omics' disciplines, biotechnology, and artificial intelligence innovation, especially in the fields of computational biology and bioinformatics, will pave the way toward a truly personalized medicine in genomic diagnostics. Integrating large data via comprehensive, personable systems into personalized health decisions could fundamentally change health behaviors, enabling precision health on all levels of health care: prevention, detection, treatment and follow-up. Anticipating that truly patient-centered genomic diagnostics will be available in the near future, individual people will have to address how aware they wish to become about body and health.

Keywords: Genomic Diagnostics, Clinical Decision-Making, Genome Alterations, Health Data, Decision-Support Systems, Data Digitization, Generative AI, Explorative Analysis, Actionable Insights, Patient Behavior, Metabolomics, Proteomics, Epigenomics, Personalized Medicine, Computational Biology, Bioinformatics, Precision Health, Data Integration, Patient-Centered Care, Health Awareness.

1. Introduction

The advent of accurate and faster genomic diagnostics has ushered in a new era in personalized health care, providing crucial information for predicting the risk of disease; identifying the need for immediate treatment or preventive measures; and selecting the optimal course of action in therapeutic decision-making. Genomic diagnostics comprise not only an individual's entire saliva or blood sequence data revealing germline variations, but also RNA expression, immune repertoire, or microbiome analyses that provide additional disease-associated clues. Expecting to have a sequence read length of more than 1 million bp, long-read nanopore sequencing technology, which has recently become available at an affordable cost, is expected to be a breakthrough technology that fully reveals structural variants in an individual's genome. However, due to the resource allocations required for high-throughput sequencing, combined multi-omics profiling, and clinical bioinformatics analysis of data, genomic diagnostics are only feasible for a small number of patients in need of clinical testing. Further, the formulation of sound personalized health care decisions based on genomic diagnostics is extremely complex. When making health care decisions, one must incorporate not only genomic diagnostic data, but also other patient information—including transcriptome, microbiome, and immune repertoire statuses—and clinical evidence linking such data to patient outcomes. In addition, specific health care pathways should be selected based on the individual's genetic background, ethnicities, comorbid conditions, and social environments. Moreover, the urgent need for a sound personalized health care decision should be considered. If genomic diagnostics are employed to address the five Ws—who, what, when, where, and why—then personalized health care decisions would be simple, sensible, and practical.

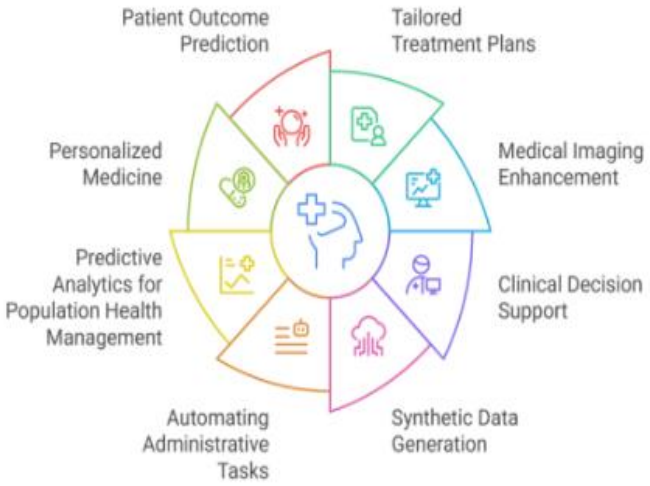

Fig 1 : Generative AI in Healthcare

The advent of molecular genetics and the completion of next-generation sequencing technology have all contributed to the rapid development and wide application of genomic diagnostics in modern medicine. Genomic diagnostics have demonstrated their advantages over traditional diagnostics in many fields, including infectious diseases, cancers, regulatory diseases, and pharmacogenomics, and have affirmatively reshaped the paradigms of clinical precision medicine. Based on current experience, genomic diagnostics are anticipated to have an even greater impact in precision health care in the future using a variety of approaches. For diagnostics at the prevention stage such as newborn screening and prenatal testing, genomic diagnostics are more accurate than other conventional strategies. They can directly check disease-causing gene mutations or even large genomic alterations which conventional technologies may miss, and with the rapid reduction in costs, genomic diagnostics for these applications are being widely utilized.

The effect of genomic diagnostics is also very important in the prediction stage, where down-syndrome prenatal diagnosis is the trail-blazing application because of the exact genetics of such a monogenic disease. For most of the hereditary diseases, however, it is not accurate enough, as a single mutation detection underdiagnosed up to 30% of the positive cases who have a known mutation in the non-tested regions. Due to the high accuracy and the comprehensive nature, NGS-based genomic sequencing have very promising potentials to be the first-line tools in down-syndrome prenatal diagnosis. For somatic pan-genomic disorders like cancers, genomic diagnostic-clinical integration has facilitated the establishment of the concept of "liquid biopsy" which is both non-invasive and repeatable. By utilizing multiple techniques to detect different levels of circulating tumor DNA, mRNA, or non-coding RNA, especially long non-coding RNA, malignant tumor development and evolution can be followed and reflected very well.

2. Understanding Genomic Diagnostics

It has long been a dream of humankind to identify the true cause of disease and tailor treatments accordingly, at least in most fields of medicine. Genomic testing paves the way for such precision medicine. Genomic testing is an umbrella term that refers to any test that analyzes DNA, RNA, chromosomes, epigenomes, or proteins. Genomic diagnostics are defined as genetic tests that look for variations in genes, including single nucleotide variations, small insertions, small deletions, larger repeat expansions, and structural variations, including large insertions, large deletions, and copy number variations, detectable by sequencing or genotyping platforms at high throughput or high coverage. Note that we define genomic diagnostics as a combination of single gene tests, multigene panel tests, exome tests, genome tests, single nucleotide polymorphism array technology, and cytogenomics with known pathogenic, likely pathogenic, and uncertain variant interpretation classification. Genomic diagnostics utilize variation detection technology, including sequencing or genotyping at high throughput or high coverage, along with crafted interpretation algorithms to seek known variant interpretation applied for use in molecular pathology laboratories for clinical diagnosis of disease. Genomic variant interpretation is informed by the claim of pathogenicity, interpretable analytic sensitivity, or analytic specificity. Genomic diagnostics pathway addresses known diagnostic biomarkers for genomic variants in disease-associated genes that provide a probabilistic basis for the use of the diagnostic biomarker analysis for clinical testing.

2.1. Overview of Genomics

Genomics is the study of the genes and their functions and related techniques, also the study of the genomes that are the complete set of genetic information of an organism. The field includes the sequencing and comparison of genomes either within or across species and the sequencing of the genomes of rare species that include microbes, plants, animals, and humans. Relying on the developments in technology, genome sequencing has become much faster and more accurate and less expensive leading to the use of genomics in every field of biology-related sciences and medicine. The various scientific sequencing technologies include microarray-based methods and short-read DNA sequencing technologies and third-generation methods. The giant amount of data generated by these technologies necessitated the need for developing

new tools and techniques for storing and analyzing these data. Genomic data analysis also includes the interpretation of the sequencing data to extract useful new biological information.

In genomics, the various phases of acquiring the useful genomic information regarding a biological system is normally referred to as, genomic data acquisition, data storage, data analysis, variant calling, annotation, and interpretation. As the tools and techniques for genomic data analysis and clinical interpretation are still under development, the move towards personalized medicine, precision medicine or translational medicine that aims is to translate the research of genomics to the clinic has a long way to go. An understanding of the next-generation sequencing methods and data is essential for the translating sequencing data to clinical practice. This chapter discusses the current state of genomic data generation and analysis and provides examples from the field of human health.

Equation 1 : Patient-Specific Genomic Signature Vector (GSV): $$\vec{G}_{\text{patient}} = \text{Encode}(G_{\text{raw}}, M_{\text{meta}}, C_{\text{clinical}})$$

Where:

- G_{raw}: Raw genomic sequences
- M_{meta}: Metadata (age, sex, ancestry)
- C_{clinical}: Clinical history

2.2. Importance of Genomic Diagnostics

Genomic information is important for achieving accurate diagnosis, prognosis, risk assessment, and treatment selection for many diseases. Although mostly used in rare diseases and infectious diseases, genomic diagnostics offer great value in oncology, pharmacogenomics, and other disease areas. In oncology, the potential for precision therapy by targeting genomic alterations in cancer has created a demand for genomic testing. The proliferation of genetic technologies has resulted in rapid reduction of the cost of testing while increasing quality and accuracy. The advent of technologies such as whole exome and whole genome sequencing now enables genomic testing to be used as first-line testing even for the most challenging clinical situations. These capabilities have made the routine use of genomic testing an expectation rather than an exception in many areas of medicine. Despite the rapid advances in genomic testing capabilities, the vast majority of physicians do not have the knowledge, training, or bandwidth to interpret and analyze sequencing data. In addition, the number and complexity of the potential number of observed variants and the interpretation problem necessitate the development of tools to help physicians arrive at a concise list of prioritized variants with associated information that can be used to make genomic-informed decisions. This need is especially acute in areas that can benefit from genomic testing capabilities, but where the clinical adoption has lagged despite existing guidelines due to ambiguity in the rules that govern the decision-making process. Genomic diagnostics enables increased understanding and better treatment selection for a beautiful bouquet of diseases that have previously never had a suitable therapeutic solution. Therefore, the coupling of ever-growing massive genomic data with state-of-the-art machine learning and generative artificial intelligence tools can accelerate the process of increasing clinical adoption.

3. The Role of Big Data in Health Care

1. Defining Big Data in Health Care

In the last two decades, rapid advances in technology to collect, analyze and utilize information have made possible the generation of large amounts of complex data, referred to as big data. Systems that handle big data, characterized by high volume, high velocity, and/or high variety, require specialized hardware and software and advanced statistical and computational capabilities. Various industries are taking advantage of big data analytics; however, businesses in the health care industry are lagging behind, in great part because of the highly complex, fragmented, and unorganized structure of the health care system. Nevertheless, in recent years, the increasing digitization of the health care industry is leading to gradients of success implementing big data solutions. Optimization of big data resources in health care is needed to realize the dream of personalized, precision medicine that takes into account unique patient characteristics and leverages the advances in biotechnology, such as genomic pathology and radiology, and drug design and development.

2. Sources of Big Data in Genomics

Genomic diagnostics is one of the major fields in health care that stands to benefit from the implementation of big data solutions. Genomic big data is in part generated from the decline in sequencing costs just 20 years after the publication of the first draft of the human genome with approximately 3 billion base pairs. The cost of sequencing a human genome has dropped to the point that today, tens of millions of genomes are estimated to have been successfully sequenced, creating a rich database of genomic information. Besides first-hand genomic data, an extensive amount of medical data related to patient diseases and phenotypic characteristics attributed to genomic variations is being generated. These data have been integrated into especially centralized large biobanks. Lastly, advances in artificial intelligence are now enabling novel technologies to predict the burden and types of diseases for patients based on their genomic variations, creating additional resources of big data.

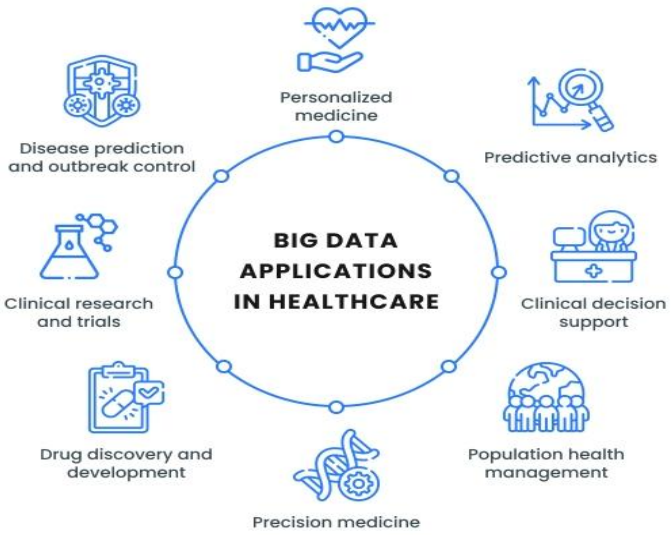

Fig 2 : Big Data Applications in Healthcare

3.1. Defining Big Data in Health Care

The concept of Big Data has been applied in diverse environments, with the most widespread use in business, technology, and governance. The emergence of Big Data in health care has proven life changing by allowing access to unprecedented knowledge from populations and by unlocking powerful new methods for population management, insight generation, and health care delivery. These new capabilities are transforming health care from a reactive model that treats disease after it occurs to a proactive model that focuses on disease prevention and population risk management. Their use is efficient, scalable, and flexible, minimizing both the cost of investment in the required platforms, and the risk of multiple adaptations to the particular needs of a specific health care application.

Nevertheless, as data sources are more readily available to a wider audience, misunderstanding arises concerning what Big Data is, and represents, in the health care space. Potential uses can range from simple correlation studies using traditional methods applied to large datasets, to the enormous computational and financial investment required for a cloud-based artificial intelligence solution capable of massive predictive analytics. These ambiguous definitions limit the capacity for investment in useful Big Data analytics infrastructures that accelerate research in health care innovation. Accordingly, we present here a definition of Big Data in health care that imposes both size and solution requirements. Both the Big and the Data aspects of our definition must be fulfilled for data and its analysis to be considered actually Big Data.

Herein, we define Big Data in health care as any health care data ecosystem that is large enough to require a specialized Big Data analytical solution. Substantial investments are the most obvious requirement. Computational infrastructure that allows organizations to house large datasets and apply specialized cloud-based services for storage, processing, and advanced analytics expands the capacity for investment in innovation for Big Data infrastructures and analytical solutions.

3.2. Sources of Big Data in Genomics

The genomic research is generating very large amounts of often shared or otherwise publicly available data. Since the advent of next-generation sequencing technologies, there has been an exponential increase in the amount of genomic data produced. Next-generation sequencing has enabled cost-effective high-throughput sequencing and a diverse set of applications in genomic medicine, such as genome sequencing, transcriptome sequencing, epigenome sequencing, exome sequencing, genome-wide association studies, and other functional genomic studies. However, challenging limitations are associated with the use of WGA of small biological material and the comparative disadvantages of some NGS technologies.

The rapid generation of genomic data from a variety of sources has also created a new concern. There are diminishing returns on spending money but not time on new NGS technologies. While most of the latest sequencing devices have now implemented protocols for generating smaller data files and data processing pipelines, they still trail in comparison to older NGS-specific data types for speed and economy. These include exome capture array kits from a variety of commercial vendors that are also optimized for and focus on large-scale population genomic studies. Other types of data in very large volumes are the publicly available reference genomes, as well as the large-scale tumor-normal DNA RNA long-read databases and image feature data from cancer studies, especially in pediatric tumors and other diseases.

3.3. Challenges in Managing Big Data

With the increasing complexity and diversity of data in genomic medicine, the challenge of big data is transforming genomic data from being merely abundant into more qualitative and valuable information. The collection, generation, and curation of meaningful data is bound to have a clinical impact only when these processes can act in synergy with the optimized computational pipelines that convert big data into actionable, clinically-relevant knowledge. On the contrary, if data generation is disordered and computational pipelines remain unsupervised and poorly optimized, big data will

likely remain a powerful tool with little or no clinical value. A sufficient degree of data generation, processing, and curation will, therefore, be fundamental to boost the establishment of robust and standardized bioinformatics pipelines able to accomplish the systematic translation of big data into knowledge and clinical actionability. Explosive technological advancement in data science and management techniques is not sufficient to guarantee the implementation of genomic medicine in the everyday clinical practice. Currently available algorithms and optimization techniques are seldom validated for their performance on elaborate patient data paved with uncertainty and errors and, where assessments have been made, the success rates have generally been poor with high variability across different groups. Moreover, much of the available methodologies have not yet been rigorously validated in the context of assessing health risks or predicting disease nor, when applicable, built into systematic clinical decision support systems. Therefore, the management of big data in the context of health care will pose challenges for both researchers and clinicians and, especially in the former capacity, fail to provide the expected level of reliability and actionability if not accurately conceived and implemented.

4. Generative Artificial Intelligence in Health Care

Generative AI has emerged as one of the most impactful computational techniques, incorporating advanced systems for generating high-dimensional data as contributions to an exponentially increasing body of AI models that are transforming numerous disciplines. Health care and biomedical informatics are seeing enhanced detection, annotation, and prediction tools applicable to a wide variety of data, including text, images, and clinical decision-making. These models, many with billions of parameters, sometimes referred to as foundation models, are having a transformative impact on therapeutics, pharmacovigilance, genomic modeling, and electronic health information. These foundation models are especially impactful along with domain-specific tuning, transfer learning, and few-shot transfer as applied to particular biomedical datasets. Recently developed models are a leading example of a foundation model for health care.

Foundational large language models and AI-enhanced multimodal models with transfer techniques or using domain-specific additional training are transforming multiple data types in biomedical fields, particularly in genomics. A variety of tools are beginning to appear for laboratory exploration and bioresearch supported by multimodal embedding for visual, genomic, and clinical information. Generative AI enhances biomedical exploration and, ultimately, models of health and disease, therapeutics, integrative functional genomics, electronic health records, or associations supported by multimodal data of multiple types. There are numerous possible areas of applications, including the data pathways of genome-to-phenome with the potential of pharmacogenomics along with health equity, building the predictive platforms. These transformative techniques and rapidly evolving capabilities of Generative AI raise numerous ethical issues around unintended biases, potentially harmful errors in prediction, generation, and embedding, privacy, and trust.

4.1. Introduction to Generative AI

Generative artificial intelligence (GAI) refers to the use of computer algorithms and systems to create entirely new outputs that mimic the intricate aspects of the material world. Recently introduced hardware and software improvements, including large-scale data utilization, powerful computing systems, and advanced algorithmic advancement, have made GAI extremely popular. Recent models have captured widespread attention for the apparent artistic abilities of the generated results. These models can generate high-resolution images, ultra-realistic digital art, websites, computer codes, text stories, news articles, poems, and blogs. As GAI-enabled tools become available, both creative professionals and amateur artists have expressed profound fatigue, discomfort, and even have concerns about potential job displacement due to the proliferation of AI-generated art. Nonetheless, with the explosive development of large and sophisticated GAI language models, the tremendous potential of GAI in producing stimulating art and creative works cannot be neglected. More importantly, despite these concerns, the potential applications of GAI go far beyond merely copying creative expressions from the realm of art, dramatically impacting many practical aspects of our daily routines.

Generative AI can be utilized in nearly all domains of artificial intelligence. Instead of only recognizing and segmenting existing objects, GAI models can create entirely new outputs and mimic material world contemplations. With foundations in computer science, informatics, mathematics, neuroscience, and robotics, GAI is a branch of AI that creates analogues of objects, images, sounds, text, and other content by simulating deep, latent structures in data; through creative imbuement, these models can expand our understanding of generative processes and co-create instruments for art and design in ways that were not previously possible. With the rapid technological advancements of AI, the GAI-enabled tools and models are rapidly transforming these domains as well.

4.2. Applications of Generative AI in Genomics

Generative AI-based approaches have, until now, been less frequent in genomics than in areas such as language and vision, a situation that may change rapidly. The use of large language models to assist in tasks such as genome interpretation annotation or increased accuracy in gene identification is becoming more common. Data-efficient approaches such as prompt tuning have been shown to be effective in bioinformatics tasks, including sequence labeling. Autoencoding models trained on contrastive objectives to replace masked RNA and DNA sequences are also boosting performance in gene identification tasks such as minimal RNA sequence saturation, homology model generation, and detection of variant effects on RNA.

Generative AI approaches based on diffusion generative models and GANs have been more common in genomics for de novo genome assembly, epigenomic data modeling, and RNA and DNA sequence generation. Innovations relative to diffusion models, designed for visual domains, include a multi-channel multimodal conditional diffusion framework for

learning epigenomic signals, and generative adversarial networks for generating high-resolution 3D genome structures from 2D data. Other GAN and diffusion-based methods include those that create chromatin accessibility maps, and those that generate DNA and RNA sequences.

Practical suggestions for scientists interested in employing GANs for biomedical data include starting with simple GAN configurations, achieving acceptable results, and then gradually increasing the model's refinement and efficiency. Guidance on the basics of diffusion models for RNA and DNA sequence generation is also available. Accessible tools for RNA and DNA sequence generation are best suited for scientists specializing in experimental wet bench work, but easy-to-use packages for analyzing bulk RNA sequencing data are emerging, driven by academic researchers, for those without programming skills.

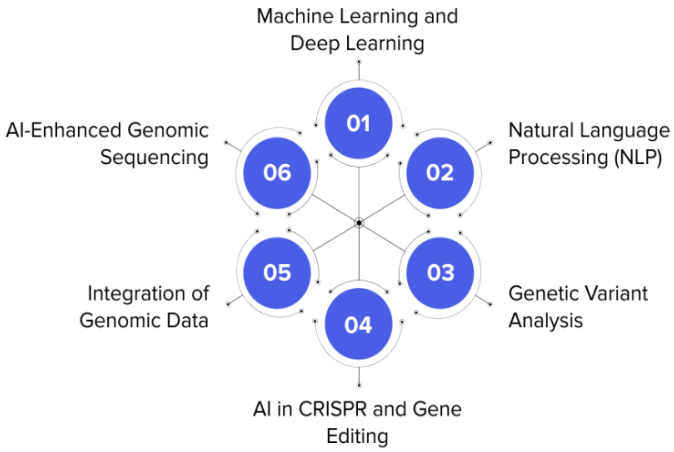

Fig 3 : AI in Genomics is Transforming Healthcare

4.3. Ethical Considerations of AI in Healthcare

Artificial intelligence combined with big data scoring have expedited the generation and subsequent exploitation of health-related prediction models and personal risk scores. Akin to being 'equally intelligent' as the user of a wizard, and 'cheaply clever' in regard to placement of tasks, these wizard tools are 'both a blessing and an insightful pondering'. Within the healthcare domain, there are several tasks wherein people might prefer to enlist AI-based tools owned and operated by the interests of a healthcare institution or commercial entity. All organizations that manage healthcare data or use predictive tools are charged with the responsibility of care, beneficence, justice, and non-maleficence towards those whose data they possess or who are potentially affected by their predictive tools. Generative tools can exacerbate things, as these tools are now considering those values as inputs and trying to optimize them as outputs for the audience of the intended user agent. The biggest debate revolves around the ability to utilize AI-generated content while listening to the social elements of the bias-variance tradeoff of the user. Because of the legacy of the algorithmic communities irresponsibly ignoring bias and considering variance the enemy, it is ethical to require AI alignment and consideration of fairness be core tenants of any organization operating advanced wizard-like tools that interface with the general populace.

5. Integrating Big Data and AI in Genomic Diagnostics

Big Data has increasingly become an empowering component in a wide range of genomic research fields and activities, driving the successful growth of a variety of application sectors heavily relying on genomic data, such as precision health. Genomic diagnostics as an active integration of genomics and diagnosis enables accurate discovery of health conditions in development or lifestyle-related diseases. Genomic data obtained through high-throughput sequencing renders diagnostic capability more efficient and effective by allowing more comprehensive examination of somatic variants and germline polymorphisms associated with disease development or progression. Artificial Intelligence, particularly, Generative Artificial Intelligence, significantly expands the applications and capabilities of Big Data. As the latest major trend in Big Data, AI will connect disparate data pieces together to make datasets smarter, to generate data-driven knowledge, and to draw further conclusions from data rather than just providing raw data support. Hence, a strategic integration of Big Data and AI in genomic diagnostics would synergistically optimize the advantages of both Big Data and AI.

Enabled by Big Data and AI, genomic diagnostics can utilize and analyze multidisciplinary data beyond genomic data alone, to better characterize the health conditions from multiple angles, and to provide more personalized, accurate, and efficient genomic diagnostic services to patients. Various efforts have been made to achieve this promising optimization by connecting Big Data, AI, and genomic diagnostics together, covering the application domains, technical components, and model configurations, and showing the performance improvement on genomic diagnostic tasks. Moreover, with the recent boom of large language models in the AI field, this integration will be much more convenient and powerful. In the following subsections, we will walk you through the concepts and successful case studies of applying AI-enhanced Big Data into genomic diagnostics, in depth, from the perspective of Big Data.

Equation 2 : Diagnostic Prediction $P(D) = \mathcal{G}(\vec{G})$

- $P(D)$: Diagnosis probability
- $\mathcal{G}$: Generative AI model

5.1. Synergistic Effects of Big Data and AI

Although the values of big data and artificial intelligence (AI) have been separately described, application as a holistic decision-making framework is less commonly described. A sufficient volume of data is the essence of AI and its applied machine learning (ML) self-correction process. Big data, particularly in omic solutions, is generated from measuring a large number and diversity of relevant elements for a particular sample type. However, an adequate number of metrics of meaningful importance for obtaining an actionable result is key. In the case of using AI for omic working solutions—predicting outcome probabilities or actions without needing to understand the complex relationship with input data—interpreting the algorithms may not be necessary. In the case of needing to explain the information from the model, global or local explanations can be obtained.

Synergy in focusing on bias presented in big data for AI—removing bad data or using methods to identify the conditions for data to be valid—can improve AI performance. Smaller, cleaner datasets can improve AIs applicability in ascertaining accuracy metrics from comparative methods. Even if there are no improvements from ML adaptation or application, improved global and local interpretability can provide important insights and validations that could stingily adapt. Big data at times is vast with differing features that cause bias. Employing techniques for grouping similar data to match with AI feature layout should increase validity. Removing or personalizing controlling types of data can usually apply for greater volume with a smaller number of features.

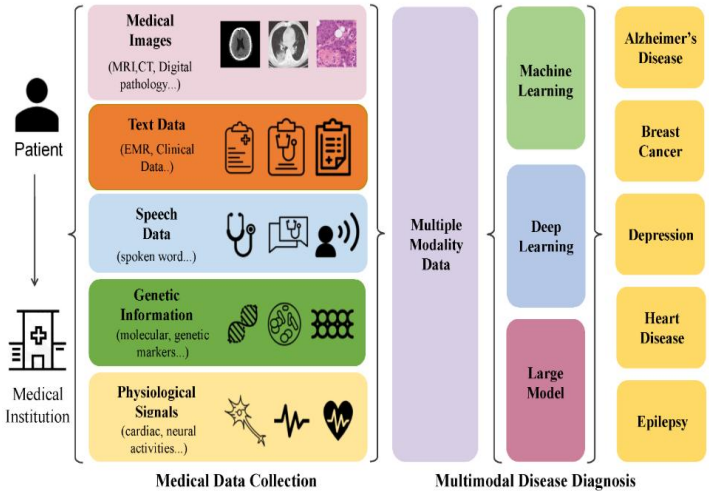

Fig 4 : Synergy of Multi-Modal Data and AI Technologies in Medical Diagnosis

5.2. Case Studies of Successful Integration

How can the underlying data science, methodology, and technological concepts behind using big data and AI together in genomic healthcare be applied in practice? Are there real-life industry case examples that show the potential of this integration? To help answer these questions, we have identified case studies from our experience in applying big data analytics and generative AI systems using cloud computing in real life. These case studies show how new generations of big data analytics and generative AI designed for genomic medicine can enable personalized, fast, efficient, and accurate genomic diagnostics, targeted and stratified prevention plans, as well as individualized, enhanced treatment recommendations and therapeutic measures. The lessons learned from both case study implementations can provide the genomic healthcare community with useful knowledge on the essential core processes and the data science and technological infrastructure needed to use together big data analytics and generative AI successfully to their full potential. The first proof-of-concept case example is the personalized genomic diagnostics for somatic mutation-driven clinical cancer subtypes. On the big data analytics side, the actual somatic mutation for the targeted oncogenic pathways and the functional annotation of the somatic mutation found in the whole genome-based integrated cancer somatic mutation data are compared against an established curated database of somatic mutation functional assessment methods. On the generative AI side, the DNA repair model is designed using the generalized Schrodinger heterogeneous mean-field model. The DNA repair pathway is implemented into the coupled general second order model to compare against.[1]

6. Personalized Health Care Decisions

1. Defining Personalized Health Care

The term personalized health care refers to tailored treatments and care models uniquely developed for patients based on various factors, such as a person's underlying biological, clinical, environmental, social characteristics or preferences. Personalized health care aims to provide unique and customized treatment strategies to individual patients while promoting

healthy behavior and disease prevention. While directed toward patients, personalized health care also considers the patient's context, family history, and societal and psychological implications of the disease and its treatment. Providing care precisely and wisely is not only beneficial to individual patients, but also helps develop appropriate care systems, willfully utilizing available resources for the improvement of public health; reducing the total cost of care, reducing the number of hospital visits, and alleviating the ineffective use of resources; and preventing and managing chronic diseases.

2. Benefits of Personalization in Treatment

Overall, personalized health care solutions can revolutionize a patient's health management and directly improve patient adherence and thereby outcomes. Personalized health care allows identification of high risk patients to apply intensively managed behavioral and pharmacotherapeutic interventions; selection of preventive strategies using the latest technology to identify patients at risk who could benefit from prevention; maximum disease prevention; the possibility to revolutionize disease treatment to convert the focus from waiting for disease symptoms until providing maintenance therapy, dysfunction management, disability support, and costly surgery and rehabilitation to active monitoring and disease prevention; avoid the trial-and-fail treadmill for medicines, to maximize benefits and minimize adverse consequences; interactive engagement, virtual support, timely monitoring; effective feedback using remote monitoring devices; leveraging artificial intelligence algorithms to help sort through large volumes of data; and integrating and coordinating care.

3. Patient-Centric Approaches

Various themes emerge from discussing personalized health care within the context of patient-centric attitudes. They include patient preparedness and preference assessment, shared decision-making, clinician-patient communication, patient engagement, treatment adherence, patient-reported outcomes, patient-centered value-driven outcomes, integrated behavioral, functional, emotional, financial, and personalized problems and solutions perspectives, end-of-life discussions, palliative care, patient-centered outcomes, and transparency. By explicitly addressing all stakeholders, including patients, family members, reimbursement agencies, and payers, guidance can be created on how to optimally measure and structure patient-centricity within the personalized health care context.

6.1. Defining Personalized Health Care

The concept of personalized health care (PHC), also known as precision medicine, was officially defined in a report that the administration hoped would spearhead a new, transformative vision usher in a decade of science, technology and discovery in diseases ranging from cancer to diabetes to rare diseases. Although personalized medicine uses traditional biomedical tools of science and is therefore not new, due to the magnitude of the acceleration of the biomedical discovery process and how personalized medicine will be informing and prioritizing selection of how and what technologies and industries are shaped. Personalized health care has been broadly defined as an emerging area of science, technology, industry and discovery that integrates novel data and technologies to improve diagnosis and outcomes of therapy and disease prevention, with an emphasis on helping implement and disseminate complex biomedical discoveries into the practical setting of patients, families, and populations facing education, social, emotional, behavioral, environmental, access, planning, and treatment decision challenges, and competing life priorities. Personalized medicine employs a new biomedical model of both science and practice of using individualized data and innovative decision-making algorithms to optimize diagnostic testing, treatment, and education, which are now possible due to advancements in our understanding of complex biological determinants, the accelerating power of novel data, tools, and technologies, and the rise of enabling industries such as information technology, data science, computation, advanced algorithms, and artificial intelligence.

The approach taken in the PHC endeavor and discussed here, is the complete integration of patient-centered care with high technology in order to optimize decision-making. These tools include whole genome sequencing, advanced computational algorithms for variant classification, targeted clinical use of next generation sequencing and other “omics” technologies. These advances help solve the “test smarter” dilemma of personalized medicine, and machine learning or other advanced algorithms driven by big data, thereby leveraging both internal medical data and data from other diverse informational sources or “knowledge curation”, which will help solve the “treat smarter” challenge. Personalized health care balances high technology with a patient-centered approach to care.[2]

6.2. Benefits of Personalization in Treatment

The traditional medicine model is based on treatments that visit the patient regardless of their identity. While the clinical practice has undergone a remarkable evolution in terms of different diagnostic tests that support physicians in their decisions, technological advances will not change this traditional model until the development of patient-specific drugs is possible. During the last years, the use of drugs targeting personalized therapies in certain diseases, such as cancer, have demonstrated an improvement in terms of efficacy and a decrease in adverse effects when compared to more traditional therapies. This is due to the fact that these therapies are studied and developed with the aim of only helping certain subsets of patients. There are also examples where non-specific therapies were effective for some patients, but the majority of respondents had considerable side effects; examples of this are certain treatments. In general terms, personalized therapies could be defined as those therapies that are supported by evidence-based medicine in their development, or are used in patients who are selected based on some specific knowledge regarding the influence of certain pharmacogenetic, genomic/proteomic, environmental, and/or lifestyle factors. In this sense, the adoption of pharmacogenetic criteria in the

development of certain drugs has increased in the last decade, mainly because the incorporation of these biomarkers in clinical trials has improved the design and robustness of the studies in development.[3]
However, the introduction of new technologies, mainly related to genomic studies that allow a thorough understanding of preventive health and disease control, have contributed to the emergence of strategies that not only identify patients who are more or less responders to a therapy but also identify the mechanisms by which the drug acts at an individual level, defining the dose and the moments for drug administration, coupling drug delivery with immunotherapies, or even eliminating the disease altogether, generating a cure. The above makes it possible to have more tailored drugs, but with a distinct objective that radically modify the traditional clinical practice, providing a quantum leap in the patient management. This radical change in the therapeutic approach in personalized medicine is possible thanks to the understanding of the entire interconnection of patient biology with disease and treatment, generating a personalized treatment plan in a move towards predictive, preventive, personalized, and participative medicine paradigm. These technologies allow the definition of more precise, safer, and more effective therapeutic strategies in terms of patient management, improving efficacy response rates and/or reducing adverse effects, as well as monitoring patient evolution more exhaustively, leading to a computed personalized support for making treatment decisions.[4]

6.3. Patient-Centric Approaches

A patient-centric health care approach for presenting and discussing the findings of genomic testing needs to take into account many factors, including the patient's precise health condition, demographic factors, and the potential for anxiety in communicating with bluntness, particularly if the discussed results are associated with risk of dying or early disability. Detailed medical history is needed to assess relevance to clinical presentation, particularly for adult-onset inherited conditions, penetrance and to establish proper controls in genotype-phenotype correlation analysis. Such history can be susceptible to social or psychological issues. An inclusion of a spouse or other close relatives in the discussion may help in more accurately obtaining the family history data.
The patient's cultural background may shape their perception of genetic testing, the significance of particular gene risk variants, and the degree of their involvement in the decision-making process, including preparatory counseling in case of testing for carrier status of applicable medically relevant conditions. The availability of targeted therapies, particularly if life-saving, may greatly facilitate discussion of even highly penetrant gene variants in severe medically relevant conditions discovered in healthy patients or carrier status of highly penetrant dominant-negative variants. When considering testing for adult-onset conditions, the advisability of testing and the strategy of how to discuss the findings are more complex. A patient may not have a fully informed understanding of what medical genetic testing entails and particular possible outcomes, may be opposed to selecting any findings that are associated with medically relevant genes, would prefer the testing to be purely for fascination, or would prefer not to pursue genetic testing.[5]

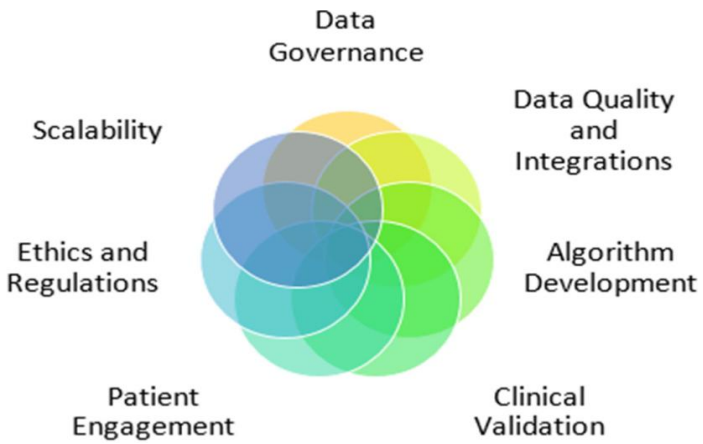

Fig 5 : Generative AI in Improving Personalized Patient Care Plans

7. Data-Driven Decision Making

Patients, their families, public health authorities, payers, pharma, and technology developers all want to make smart decisions about health and disease. Thus, it has become vital to adapt data-driven models from other domains and develop new models for evidence generation for use by those making health-related decisions. This not only requires experts in various health domains, but also statisticians, economists, social scientists, and other methodologists to collaborate to enhance the value of the vast amounts of real-life health data now available. The overall framework is relatively straightforward, though far from simple to implement. It starts with asking the question of decision relevance. This is followed by the identification, assimilation, and curation of relevant real-world health data, including the development of new digital data collection mechanisms as required. Advanced methods from AI and machine learning to agent-based modeling then analyze the data to make the best possible inference about probabilities of different potential outcomes based on different decisions.
The final steps require more collaboration among domain experts, methodologists, and those making the decisions about what information is required and how best to communicate it back to decision makers. The aim is not just a summary of

various results according to who gets the treatment, but rather relevant, tailored analysis based on simulation of diseases and of outcomes of choices using the AI-assisted computational agents. In an era of regulation of human health-related activities due to huge computing capabilities and computational costs, the time has come to maximize the still unrealized potential of the rapidly growing stores of digital information about the enormous amount of real-world information on the many choices we humans make related to our health and its outcomes.[6]

7.1. Framework for Data-Driven Decisions

Data-driven decision making is an interdisciplinary area of focused inquiry that integrates big data, AI augmentation, personalization, and behavioral economics to train decision models that accurately reflect actual decision patterns in complex environments and modify these decisions decisively to impact performance improvement metrics meaningfully. Data-driven personalized healthcare decisions harness the power of the behavioral framework. The right available data in the decision realm measures actual healthcare behavioral decision making – economic, clinical, cultural, ethical, financial, logistical, psychosocial, and societal. Data-driven personalized healthcare framework combines the science of behavioral economics with the technologies of big data and generative AI to shift the focus from care delivery on what clinicians recognize on the basis of their prior education and experiential learning, combined with state-of-the-art software tools, to what actual patients want and need, plus the triggers that will induce them to engage in these practices and make the right decisions. It considers two major enabling components: (i) improving the quality of care with respect to specialty recommendation, pre-visit behavior, quality of diagnosis and treatment, and post-visit follow-up and revision; and (ii) ensuring a positive experience with regard to diagnosing what the patient wants emotionally, culturally, and ethically, and satisfying this need empathetically and expediently.

Broadly categorized, there are three major types of data on healthcare behavior that comprise the clinical outcome data structure: (i) patient-incentivized, -driven, -organized, and -evaluated surrogate outcome metrics, i.e., time to diagnosis, time to clinical decision implementation, time to treatment initiation, time to waitlist removal, time to symptom resolution and amelioration, and time to cure; (ii) verified, -captured, -recorded, -coupled, -assembled, and -visualized diagnostic physical measures that standardize the actual healing effect, i.e., functional status, cure rate, symptom alleviation and resolution, complications and from procedure and treatment; and (iii) fully-session rated, -assessed, -collected, -stored, and -indexed qualitative psychosocial assessments that address subtle “dependent” and complex private feelings patients experience through their healthcare journey and process. We believe that this composite patient data enables healthcare decision-enhancing personalization.[7]

7.2. Impact on Clinical Outcomes

Data-driven decision support will surely have its impact on clinical outcomes. Improvements to diagnostic processes, patient stratification, personalized therapeutic approaches, and better assessment of drug efficacy and toxicity will directly tie results from DDDM to tangible patient benefits. Improved diagnosis capabilities could result from faster turnaround times, more reliable detection of pathogenic alterations, and the ability to measure actual tumor dynamics through longitudinal plasma analysis rather than static cross-sectional tissue snapshots. Moreover, DDDM could additionally offer a risk-benefit assessment for rare diseases, assessing the possibility of finding an identified genotype for the patient and the pros and cons of a successful or unsuccessful therapy response reduction. Indeed, DDDM is very much needed in the treatment of cancer, CNS disorders, and rare diseases. The difficulties presented by these diseases can benefit from advanced machine-learning models that can crunch large amounts of data across the different existing cohorts, including additional datasets that would initially be ignored because of differing ethnic origin, ancestry, and population heterogeneity. The increased genomic diversity, understanding of ancestry, and genetic risk factors for certain diseases will allow a better stratification in the treatment of complex diseases where all treatment responses are reduced or altered. Cancer and other complex diseases are also those fields where generative AI approaches can potentially improve existing algorithms with better feature extraction and disentangled representation.[8]

7.3. Real-World Applications

In practice, personalized health data enables physicians to customize treatment decisions for their patients based on the patients' biological profile against group population statistics. Analyzing EHRs data has improved patient care through improved decision support, assisting clinicians in adopting evidence-based guidelines in diagnosing or managing their patients' conditions. Clinical cohorts in Genomic EHRs provide realized factual information about what genomic variants, variant interactions, variant-phenotype relationships, and pathways lead to which medical conditions are likely to affect clinical outcomes, and are associated with drug responses or adverse drug reactions. That invariably supports patient-specific and rational patient-priority manual perusal of genomic and associated phenotype data for developing diagnostic or treatment plans for patients with specific conditions, to ultimately improve clinical outcomes.

We have also developed data-driven clinical databases addressing specific clinical problems leading to impactful results. The GIANT portal consists of large-scale diverse genomic and gastrointestinal phenotype data of patients and controls that support data-driven inference of GI phenotype-genomic associations for decision support in patients with GI conditions, and offer clinical clues for treatment response in patients who receive GI-directed treatment. The human immune system is home to unique cellular gene expression programs deciding developmental and functional plasticity of major immune cell lineages and state in humans. The IEMDB is a large-scale interactive database containing data-driven

CD4, CD8, Treg, B, NK, and monocyte-macrophage cell type-level unique gene expression programs, and program-based immune cell network interactions across human organs.[9]

8. Future Trends in Genomic Diagnostics

As technological advancements continue along the exponential growth path that has defined the past two decades, a number of constraints that today limit the scope of genomic diagnostics will be erased. These advancements will serve to enable new applications and new service delivery techniques that will transform healthcare into a data-driven and value-based industry. Miniaturized diagnostics are already being embedded as wearable devices or chips that can be deployed in the field. New methods for the automated analysis of sequencing data are rapidly being developed, enabling real-time in-field sequencing of biological agents.

The implications of this type of technology development for genomic diagnostics are vast. AI developmental advances will make it possible for genomic diagnostic assessments to be done in real-time, opening up a rich set of novel applications in healthcare applications such as surgical planning/guidance, emergency triaging, remote personalized care, and primary care addressing physical and mental well-being. These will further free primary care physicians to focus on the complex cases requiring their expertise, while maintaining high telehealth utilization rates and personal engagement. Adoption of robotic implantable nanoscale diagnostic probes will enable ultra-high temporal resolution diagnosis of a multitude of pathological conditions in real-time. The deployment of liquid biopsies for the assessment of tumor burden through the number of circulating tumor cells or the level of tumor exosome concentration will make it feasible to actively monitor responses to treatment, and identify recurrences well in advance of the reappearance of clinical symptoms. Residual burden following neoadjuvant approaches will be detectable and enable personalized treatment alteration decisions prior to surgical intervention. AI-based personalized risk and outcome predictive models will enable real-time clinically actionable deep-dive genomic data assemblies. These will enable the ready use of broadly facilitative health predictive models, serving to provide continuously updated predictive risk profiles, cohort membership status, and clinical actionable insights of significance to the responding individual and their healthcare providers.[10]

8.1. Emerging Technologies

Pioneering advancements in technology will bring major changes in our personal lives for caring for our health and the health of our family members. The growing popularity of consumer and clinical genetic testing and the availability of many peer-to-peer genomics applications is heightened by the wide net adoption of smartphones and mobile clinical services in the last decades across the globe. Along with predictive and preventive medicine, these developments will open multiple possibilities for the delivery of personalized care. The creation of joined medical genomic databases in the cloud wherever personal as well as family genomic information is shared will facilitate large-scale studies exploring lifelong health trajectories. The advances in deep learning algorithms for data organization and preparation and the emergence of cloud neural network processing will aid in the identification of rare or latent gene interactions with health issues. Distribution of handheld next-generation sequencing devices, AI-generated, long synthetic-genome sequence data, and digital twins can change the way precision health is tracked and enhance the availability of predictive algorithms to recommend early action.

Translational medicine increasingly depends on the application of advanced technologies for the generation of data in nano or low volume. In this chapter, we discuss processes affecting the field of NGS and the identification of genomic variants using AI-derived discovery architectures capable of assembling complete genomes and explore AI use cases that represent emerging enabling technologies likely to be integrated with NGS in health and wellness tracking. NGS platforms will become smaller in size and lower in cost without compromising sensitivity, specificity, and dynamic range of performance. Devices will be incorporated into tools for use in mobile detection of variants of interest or in triaging or ruling out targeted conditions in patients of genome stratified at risk. Whole genomes will be sequenced.[11]

8.2. Potential Impact of AI Advancements

Advancements in generative AI and multimodal transformers have the potential to deliver stunning progress in the field of genomic diagnostics. The rise of these technologies can be compared to the leap made in computer vision in 2012, when a significant increase in image classification accuracy enabled by deep learning was demonstrated. Innovation and development of new tasks, such as Zero-Shot Classification, was propelled by the introduction of large image classification models and fueled enthusiasm for deploying AI technology in multiple different industries and specific applications. Just a few large language models have achieved human-level performance in text-based classification tasks, enabling implementation of practical applications that were widely adopted by individuals and companies. Prediction of the genomics progress curve is less reliable than prediction of computer vision because of the particularities of the biology field, but relatively easy tasks, such as simpler genomic variant detection and effect prediction, represent low-hanging AI fruit.[12]

Utilization of AI technology in genomics field applications, particularly in the area of clinical diagnostics and decision-making, will become possible when human-level performance is attained for specific healthcare and genomic tasks. Accelerating the pace of innovation in diagnostic tasks carried out by AI models in genomic diagnostics most likely will require collaboration between model developers and subject matter experts on implementing models in real-world applications. Improved accuracy and better interpretability of AI models for decision support in genomic diagnostics systems will enable the scaling of clinical genomic applications from genomics-native companies to mass-market adoption

by hospitals and diagnostic laboratories. These systems will enhance speed and precision in handling both expert and consumer genomics testing options and improve outcomes of cancer prevention and accelerated precision therapy selection. The transition from the era of laboratory technologists performing the majority of the genomics tasks to bioinformaticians and clinicians leveraging AI for analysis of testing data will happen as soon as the task performance of models clearly exceeds that of human experts.

Equation 3 : Treatment Decision

$$T^* = \arg\max_{t \in \mathcal{T}} U(t)$$

- T^*: Optimal treatment
- $\mathcal{T}$: Treatment set
- $U(t)$: Predicted utility of treatment

9. Challenges and Limitations

Generative AI has shown promise for personalized health care decisions, especially those powered by big data. However, there remain challenges before it can be widely deployed, particularly in diagnosing genetic diseases. These challenges include those inherent to machine learning (ML)-based systems, such as biased training data that induces biased results, issues with interpretation of rich output data, and the quality of generated data, including inaccurate or nonsensical results. In addition, there are important regulatory, compliance, and privacy concerns associated with deploying a generative AI model in the healthcare space.

Arguably, the above issues are less about generative AI and more about machine learning in general. For example, many of the technical limitations discussed are not unique to generative AI, but common to all ML-based systems. There exist toolchains and other strategies to deal with ML issues, including mitigating biases, related explainability of rich outputs, and ensuring the quality of results. However, these existing strategies are not completely sufficient to deal with the increased difficulty and/or inherent uncertainty of health care applications. Generative AI is simply a more advanced computation strategy than traditional ML-based methods. However, the incorporation of generative AI into patient care decision-making promises even wider impact than traditional ML methods. While traditional prediction methods have had a large influence, personal genomic analysis and support decision-making related to those results will be substantially more impactful and beneficial at a personal level than prior ML-assisted tools. At the same time, the challenges associated with generative AI in such use cases are greater, related to the critical nature of health care functions that would be impacted by these models and tools.[13]

9.1. Technical Limitations

A number of technical limitations constrain the degree of success of personalized health care decisions powered by big data and generative AI. None are presently insurmountable, but they will add to resource requirements and/or limit the ability of the resulting analytic to meet desired performance standards. First, the data necessary for the generation and use of such clinical decision tools will typically not be homogeneous in its structure, with many different types, each with its own model, of data flowing in from different aspects of the patient's history, clinical workup, and planned treatment. In addition, the transcoding of different types of data into a common, model-independent representation may be quite complex, with many variables not captured, thereby increasing the risk of bias and amplifying any such bias already present in one or more of the data sources. These risks can be ameliorated through much experience in transcoding the variables present in both the patient history and various contributions of big data, but there is obviously a limit to the experiences accumulated that can help.

Second, where data is translated into numbers that serve as the inputs to quantitative methods, no absolute or even well-defined scale of measure is likely to be found. Many disciplines associate certain types of data with a fuzziness in measurement, such as could be found in the similarity of word meanings across documents generated in different contexts, or even badly translated from one language to another. While word embedding does provide a framework for numerical representation that is generally accepted, several aspects of fuzziness may still compromise performance. For example, element-wise distances between words may be too sensitive to very small differences in vocabularies, or each word's embedding may change substantially depending upon the document's content and word ordering. Such fuzziness may also increase the risks of not detecting and correcting for bias. Finally, the large size of the data merged for the purposes of personalized health care decisions may slow the speed of building the model and generating predictions.[14]

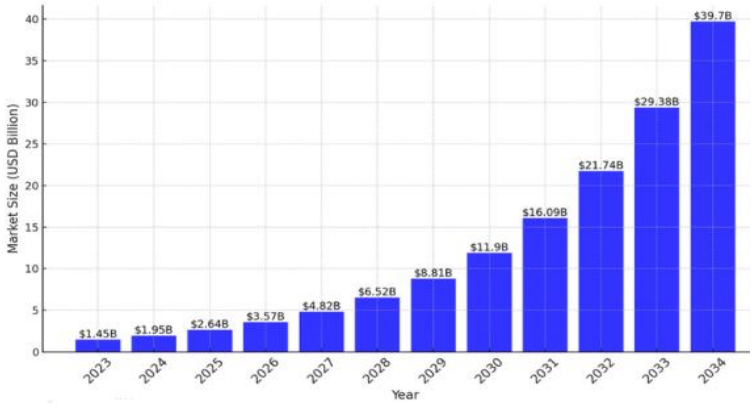

Fig 6 : Generative AI in Healthcare Market Size

9.2. Regulatory and Compliance Issues

The personalization of health care decisions, powered by generative artificial intelligence and applied to the domain of genomic medicine, indicates the need for a framework that assures participants that genetic and health data interchange is highly regulated and protected. Establishing a regulatory and compliance structure ensures health data and genetic data, the most privacy-sensitive information in existence, is properly authenticated and protected using unique identifiers for all parties involved in the data transaction. While health care patient data has been safeguarded using governmental regulations for decades, genetic data is now being utilized for several proprietary and commercial purposes, thus requiring amendments to established regulations. Organizations involved in the development and launch of GenAI applications supporting genomic medicine solutions should familiarize themselves with the requisite regulatory and compliance requirements and work closely with governmental regulatory agencies and third-party auditing companies to provide their target market with the strong confidence that their genomic data privacy and security is assured throughout the entire life cycle of the product development and deployment of the application. Regulations need to address the specific issues relating to the models associated with AI in order to ensure compliance, both through regulatory compliance by the organization developing AI and through regulatory compliance by the association supplying the data to the organization, such as a hospital, for use in generating the AI output.[15]

The unique issues around genome interpretation can mean additional regulations or amendments to existing regulatory bodies on data security and identification. While the main Act for protection of health information remains, there are extremely sensitive and nuanced amendments to consider in conjunction with other organizational and legal requirements regarding medical devices, laboratory testing, and guidelines for software as a medical device.[16]

9.3. Patient Privacy Concerns

Emerging technologies along the continuum of care can offer invaluable assistance throughout the four phases of personalized health care decision-making, namely, reporting, assessment, analysis, and interpretation. However, patients and families are increasingly aware of and sensitive to the unintended consequences of technology on their privacy. For decades, there has been an implicit social contract: access to and sharing of an individual's health information with health care providers and in some cases those who offer payment for those services are primarily responsible for making decisions about that person's treatment. Except when those decisions conflict with the decision of the individual, the professionals involved in caring for that person have unrestricted access to that individual's health data. The patient risks compromise to their physical well being through their data files in exchange for access to the best possible care.

Generative AI applications create data by generating information based on prompts from users. In the case of conversational models, that interaction is predominantly with human beings with human-to-AI chat systems or social media-based chatbots being relatively new entrants into the field. Individuals create the data made available to the companies operating these generative apps; however, that data is often monetized by developers to build their platforms, which enables them to improve upon the information users of their products provide. Ethics surrounding big data and specifically generative artificial intelligence are at the forefront of debate, and questions have been raised regarding how health care professionals should utilize such resources to make better decisions for their patients while minimizing the risks. This chapter evaluates both the opportunities and the risks.[17]

10. Conclusion

In this paper, we presented the innovative application of generative artificial intelligence for patient education and clinician decision-support in genomic diagnostics decision-making. The emergence of big data and generative artificial intelligence technologies has eased important limitations of prior applications of artificial intelligence in genomic research and clinical medicine in personalized health care that involved relatively small datasets, lack of deep technology integration such as for employing natural language processing and neuro-symbolic methods, and lack of real-time decision-support and patient and clinician engagement. Our proposed Genomic Decision-Support and Clinical Guidance Algorithms including 1) patient education capabilities via patient questions-answering middleware tools, 2) clinician decision-support via generative artificial intelligence-assisted clinical workbench for analyzing genomic diagnosis platforms and determining

the optimal platform for patients with presumed Mendelian conditions, and 3) patient-centered generative artificial intelligence-assisted clinical decision support that jointly considers patients' preferences and both technical and clinical considerations of genomic diagnosis, as well as employing a platform for sharing genomic data to speed the diagnosis and discovery stages of precision medicine, are examples of integrating generative artificial intelligence, multimodal big data, and existing multi omic, phenomics, and clinicomic data and analyses. These capabilities will contribute to optimizing the decision-making around the genomic diagnosis process, thereby enhancing precision medicine by expediting the genomic diagnosis process and optimizing the use of genomic technologies.

There are several future directions for research as our preliminary focus has been primarily on the educational and decision-supporting aspects for the genomic diagnostics process. Increasing patient, documentary, imaging, multi omic, phenomic, and sociocultural diversity while recognizing the effects of charting disciplines and domains in the Clinicomic domain would enhance the multimodal big data input for the generative artificial intelligence models, and possibly resulting in better performance of these models across demographic groups and multiple diagnostic use cases. Such data may reduce the chances of failures, biases, and health care disparities associated with bad actors perpetrating biased artificial intelligence Genomic Ethics violations while enabling the virtuous cycle of enabling and democratizing personalized and precision medicine possible through generative artificial intelligence and big data in genomic diagnostics in an increasing proportion of patients with presumed Mendelian conditions in a patient-centered manner.[18]

10.1. Final Thoughts and Future Directions

Advancements in big data, generative artificial intelligence technology, and bioinformatics have paved the way for personalized health care decisions powered by big data and generative AI in genomic diagnostics. Personalized decisions regarding what genetic variant to report back to patients and their family members are supported by real-world experience and populations, deep generative models, and ontology representational learning. This framework minimizes the distance between genetic variants and their semantically similar variants in reports. We developed bioinformatics and generative AI technology tools to facilitate this novel personalized decision-making framework in genomic diagnostics.

The present study proposes future directions in advancing the era of precision medicine using the personalized health care decision schema for histopathological and genomic traits: Generally accepted that a deep learning consortium can add value to the histology diagnosis of cancers. The next step is to combine genomic data with histopathological data using generative AI. This project aims to genetically explain the histology of every tumor in the world. A collaborative effort could lead to crowdsourcing histopathological data to develop an easily accessible real-world population repository. Genomic data from patients with common or rare diseases and their healthy family members from around the world must all be deposited into one integrated and anonymized repository. Together with advancing putative function prediction of variants of uncertain significance, we believe positive and negative variant associations may allow custom-built risk assessment tools to be developed. [19]

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