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Empowering Precision Medicine: The Role of Personalized Genomics in Advanced Healthcare Systems

Anju Arya*[1], Amita Malik[2]

[1]Deenbandhu Chhotu Ram University of Science and Technology, Murthal, Sonepat Haryana, India
Email ID: anjuarya12@ymail.com

[2]Deenbandhu Chhotu Ram University of Science and Technology, Murthal, Sonepat Haryana, India
Email ID: amitamalik.cse@dcrustm.org

***Corresponding author:**
Anju Arya
Email ID: anjuarya12@ymail.com

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ABSTRACT
For years, the medical field has been striving to provide personalized healthcare to patients. The traditional methods in practice are not flexible enough to provide personalized treatment to patients. Before prescribing a medication, the typical tailored method has relied on examining physical symptoms and clinical test results. Yet, the recommended therapy is often based on a "one size fits all" methodology, a solution that works for everyone in the population, despite the fact that every person's body responds to treatment differently in a diseased state. There has been a clear change in recent decades from evidence or proof-based medicine to precision medicine, which focuses on a person's traits. The personalized genomic medicine strives to address the inadequacies of traditional personalized medicine approaches, thereby complementing the precision medicine concept of selecting suitable treatment options and increasing patient response. The motive of personalized genomic medicine is to ensure that a person receives the "Right decision and Right therapy at the Right moment." In this paper, we will provide an outline of personalized genomic medicine and explore its potential in transforming the current health-care system in the near future.

Keywords: *Personalized healthcare, Precision medicine, Genomics medicine, Personalized medicine*

1. INTRODUCTION

Providing quality healthcare and treatment is the primary goal of the healthcare system. The medical science focusses on treating human ailments/diseases through biological molecules that can directly affect the afflicted organ or condition. The current healthcare system emphasizes a "one-size-fits-all" method to therapy. The therapy options for a specific disease are determined by the pathological changes and clinical symptoms. However, it has been found that a general solution that is effective for a community may not be effective for a person. Many individuals do not obtain timely and effective treatment that is appropriate for their medical condition. A drug's efficacy and side effects differ from person to person, depending on his surroundings and lifestyle [1]. For instance, [2] in the case of diabetes, 43 percent of the population does not respond to the prescribed treatment, 40 percent in the case of cardiac arrhythmias, 70 percent in the case of alzheimer's, and 75 percent in the case of cancer. There is a now a demand for a true revolution that will transform the previous "one size fits all" strategy into a more effective method. The solution is "Personalized Genomic Medicine," which treats each person as unique individual in terms of treatment and provides personalized medication [3]. It's critical to get the appropriate knowledge and approach on time if you want to get the right and early treatment, especially for lethal conditions like cancer. This goal can be achieved with the help of personalized medicine. Personalized medicine or care is not a new notion, it has existed in the health care system since 1960s [4] and gained popularity in the early 2000s [5]. The concept of personalized medicine was predicted by Sir William Osler (1849-1919) [6]. It provides patient care using genomic or non-genomic techniques for various diagnostic or therapeutic purposes [7]. Treatment with warfarin, for example, can be influenced by both hereditary (CYP2C9and VKORC1) and non-genetic variables such as sex, age, smoking, bodyweight, and drug interactions [8]. This information has the potential to change drug dosage by more than 50%. Even some cutting-edge developments in biochemical such as genotyping, single-nucleotide polymorphisms (SNPs), biochips have assisted in making personalized medicine feasible [9].

Personalized medicine, precision medicine, and personalized genomic medicine are all terms that are used interchangeably to refer to the same concept, but they have distinct meanings depending on how they are used. All of these, however, fall under the umbrella of a tailored health-care system. Personalized medicine deals with the classification of individuals into population subgroups that correspond in their vulnerability to a certain disease or their response to a particular therapy in order to tailor medical care to each patient's unique characteristics [10], [11], according to the President's Council of Advisors on Science and Technology. It aids in the selection of a treatment that is appropriate for a certain medical problem based on a person's genetic composition. It aims at avoiding trial and error phases of drug and therapies by using genetic profile of a patient to minimalize adverse effects [12]. The major objective of personalized medicine is to facilitate prevention, diagnosis and treatment of a disease [13]. Furthermore, personalized medicine does not imply the development of a novel drug or gadget tailored to a person's specific needs; rather, it merely categorizes people based on their illness susceptibility and treatment response [14]. Personalized medicine is a type of medicine that focuses on the values and viewpoints of the patient. Precision medicine refers to a strategy of adapting medical therapy to the specific factors that are unique to a person, in addition to the generic factors. It takes into account and employs information about genetic variety of an individual, his lifestyle, and the environment in which he or she lives or is exposed [15]. Precision medicine has three main goals [16]: to optimise and improve healthcare, to reveal pathogens and comprehend their mechanism, and to provide precise and effective therapy. Precision medicine, according to Edward Abrahams, President of the Personalized Medicine Coalition, is concerned with the relationship between a diagnostic test and drug use. Genomic medicine is a branch of medicine that uses genomic data to make diagnostic or therapeutic decisions in clinical care [17]. It can also influence medical decisions by using information from genomic derivatives including RNA, proteins, and metabolites [18]. Genomic medicine is a branch of medicine that applies genomic knowledge to clinical settings in order to provide more individualized care. Mammaprint, Oncotype DX, gene-expression therapy, gene replacement therapy, and other treatments are examples. Personalized genomic medicine aims to find right treatment at right time for the right patient [19]. It uses genomic tools to obtain the required genomic information, which falls under the category of modern healthcare tools. The Figure 1 has summarized different categories of Health care tools.

Personalized genome medicine and its significance is examined in the realm of personalized healthcare in this study. We shall explore genes and mutations and their relevance to a person's health state in the next section because that is the reason for the creation of this notion in the health-care system. The contribution of the paper to personalized healthcare system via incorporating genomic information can be summarized as follows:

- We highlighted the relevance and need of the personalized genomics to support personalized healthcare system.
- We have discussed the relation of gene mutation to a person's health and significance of genetic tests.
- We discussed the field of personalized genomic medicine in detail, which includes its need, challenges, and applications.
- We have discussed various trends and technologies supporting personalized medicine.

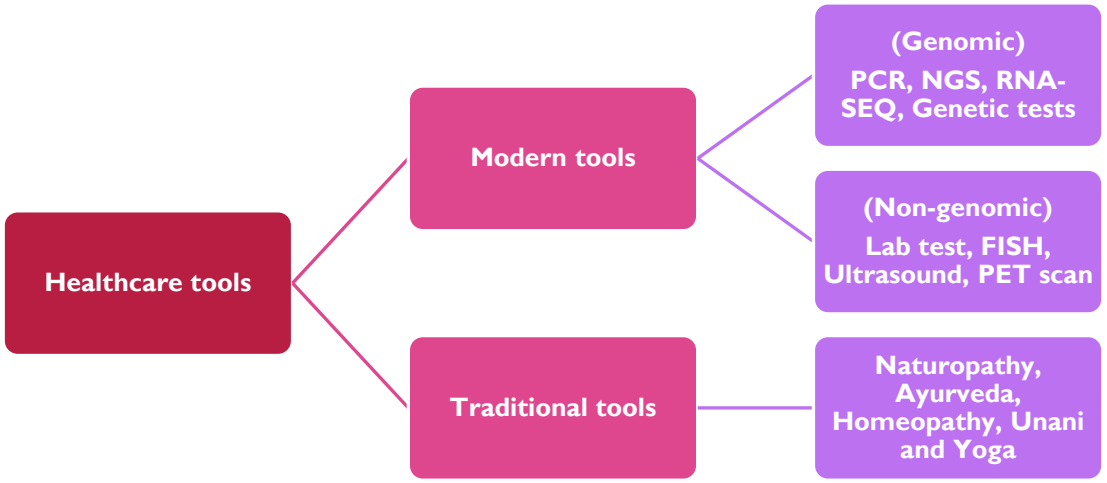

Fig 1. Categories of Healthcare tools

Paper Organization

The remainder of the paper is organized into the following sections: Section 2 provides an overview about genes and how mutations may affect a person's body; Section 3 will discuss about the rise of genomics in medicine; Section 4 provides an overview about personalized genomic medicine and then the need for genomic medicine, the benefits, challenges and application areas of personalized genomic information; In Section 5, trends and technologies that are being used to support personalized medicine is discussed; Section 6 discussed the relevance of genomic information in healthcare and Section 7 provides the concluding remarks about the significance of genomics in improving quality of life and the technologies that can support this process.

2. GENE MUTATION: NEED OF GENETIC TESTS

Genes are the fundamental unit of heredity, consisting of nucleotide sequences that carry information about a person's characteristics and health status [20]. The instructions for creating proteins that define the functioning and behaviour of the entire body are stored in genes. A human genome has around 20,000 and 30,000 genes. The gene information exposes family history, a person's health status, and risk of developing a disease in relation to a specific lifestyle and environment. A gene mutation [21] is a permanent modification or alteration in the DNA (Deoxyribonucleic acid) sequence that differs from the usual sequence present in other healthy people. Gene mutations can be categorised as Acquired mutations and Hereditary mutations (see Fig 2). Hereditary mutations are passed down from one generation to the next, but acquired mutations, which can be observed in affected body organ cells, can happen at any moment during a person's life. Internal mechanisms (such as errors in DNA copies during cell division) or external factors (such as ultraviolet radiation) might cause these mutations.

The normal functioning of the related proteins is disrupted by gene mutations, resulting in a medical disease known as genetic disorder. Not all mutations result in disease. Some mutations have no effect on a gene's ability to produce a protein. Many polymorphisms exist that have no effect on health but may influence a person's disease risk. Disease-causing mutations, on the other hand, are uncommon in the general population. Mutations can sometimes have a beneficial effect on the body, however this is a rare occurrence [22]. Specific genetic variations (inherited or acquired) cause genetic vulnerability (predisposition) and may contribute to disease development indirectly. Certain mutations in the BRCA1 or BRCA2 genes, for example, enhance an individual's risk to ovarian cancer and breast cancer. Variations in the BARD1 and BRIP1 genes, on the other hand, play a limited role in increasing the risk of breast cancer. It is not uncommon for an individual with an inherited genetic propensity to never develop an illness, despite the fact that other family members have. In the event of genetic predisposition, the chance of developing a disease depends on a variety of factors including an individual's lifestyle, environment, and other hereditary factors in multifactorial disease. Although an individual's genetic composition cannot be changed, adjustments in lifestyle and environmental conditions may reduce the chances of a disease development in persons who have a hereditary susceptibility to it.

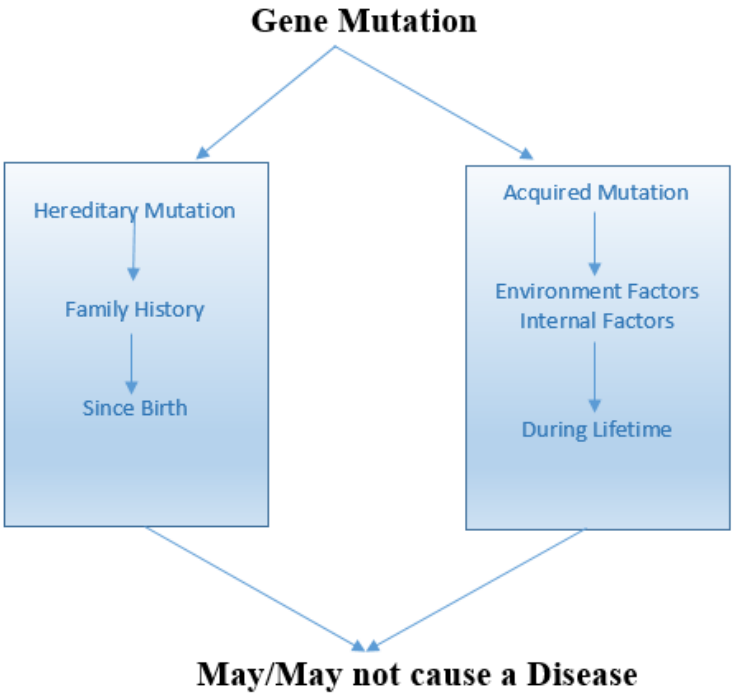

Fig 2. Categorisation of Genetic Mutation

It can be challenging to diagnose hereditary diseases. Finding a link between a genetic change and the aetiology of an illness is difficult, especially for genetic mutation or variants of unknown significance (VUS). Copy number variations (CNVs) in chromosomes, chromosomal alteration (loss/gain), and structural changes in chromosomes could all be reasons of mutations [23]. These changes may or may not cause disease, but they do have an impact on disease progression and even affect drug response.

Genetic testing is used to investigate a group of genes and identify a person's medical condition. For instance, single gene test can be performed for diseases such as Duchene muscular dystrophy or sickle cell disease and genetic panel tests for diseases such as colon cancer or breast cancer. Whole genome or DNA sequencing methods can be used to learn everything there is to know about a person's internal and external traits. GWAS (genome-wide association studies) aid in the discovery of genetic variations. The changes in genes, chromosomes, or proteins are detected through genetic testing [24]. The information gathered can help confirm the existence of a hereditary condition or predict the chance of developing one. It may also help predict the chance of a person passing on a genetic on a genetic disorder to the next generation. There are currently over 1,000 genetic tests accessible. A genetic test can cost anywhere between $100 to $2,000, depending on the type of genetic test and its complexity. Genetic tests can be performed from a variety of angles in a variety of situations, including research and therapeutic settings. Genetic testing based on research focuses on identifying unknown genes, understanding how they act, developing tests for future therapeutic application, and increasing knowledge about a genetic disorder. Clinical genetic testing is used to diagnose the cause of a condition, such as a hereditary disorder. The paper [25] presented an overview of some of the most common genetic tests and how they might be used to provide tailored therapy. Every person is genetically distinct, and susceptibility to disease differs from person to person [26]. Understanding the relationship between a gene and drug reactions and diseases aids in a more accurate assessment of hereditary risks. For monogenic illnesses, the utility of genetic tests has already been demonstrated. Thus, understanding the functioning of a person's body in a medical condition and deciding on a future course of action or therapy requires knowledge of the person's genes or genome.

3. THE ADVANCEMENT OF GENOMICS IN MEDICINE

Prior to the 2000s, genetic tests were only utilized to diagnose a few aberrant illnesses. Because of their accessibility and expense, these were not suggested for a general medical problem. However, the genomic era [27] began in 2003, when the Human Genome Project (HGP) met its objectives [28], [29]. Low-cost communication and computing technologies helped their implementation in medical at the same time. Its clinical applicability [29] was initially hinted at in 1999, when microarray-based transcriptional profiling was used to differentiate between lymphocytic leukaemia and acute myeloid leukaemia [30]. Early efforts to personalized medicine were focused on genetic variations, which helped predict drug response and prescribe dosing. One example is Phenytoin dosing in [31], which led to the development of clinical practice standards driven by the human genome. Furthermore, [32] the advancement of next-generation sequencing technology, which has resulted in lower prices, has enabled genetic data to be used to make informed therapeutic decisions.

The previous Milestones for Genome Sciences and Genomic Medicine through 2018 are discussed in a paper published in [33]. The International HapMap (Haplotype Map) Project [34], which began in October 2002 with the goal of discovering single nucleotide polymorphisms (SNPs) and copy number variations, was one of the primary projects established along the lines of HGP (CNVs). The data on these frequent genetic variants was compiled, which will aid future genome-wide association studies. The catalogue was helpful in the development of first-generation microarrays, which can assay thousands of genetic variations in a single test at a reasonable cost. The 1000 Genomes Project, a new genomic mapping consortium project, was launched in 2008 [35]. The goal of the project was to sequence the genomes of 1,000 people in order to discover the most frequent genetic mutations and compile the information into a comprehensive database that could be shared with researchers. The National Human Genome Research Institute (NHGRI)-EBI GWAS catalogue [36] includes 5687 GWAS with 71673 variant-trait associations gathered from 3567 articles for various diseases, biomarkers, and drug responses. Around 75,000 commercially available genetic assays [37] are now available commercially, increasing the use of genome sequencing for routine clinical diagnostic purposes. It is currently widely utilized in the detection and treatment of cancer [38].

Multi-omics has the potential to enhance knowledge for complex diseases, especially when integrated with other fields such as systems biology, computational biology, bioinformatics, and clinical research. Combining diverse omics data including, genomics, proteomic, transcriptomic, and metabolomic, can significantly improve patient’s treatment strategies and minimize side effects. In short, multi-omics approaches make a pivotal contribution to the advancement of personalized medicine [39].

4. PERSONALIZED GENOMIC MEDICINE

Personalized genomic medicine [7] defined as the personalization of healthcare based on a person's genetic profile. It takes into account a variety of elements such as genetic information, family history, medical history, environment, and lifestyle. PGM, often known as "P4 Medicine," is concerned with treatments that are "personalized," "predictive," "preventive," and "participatory" in nature [40] (see Fig 3). Personalized genomic medicine can reveal information on inherited diseases, rare

diseases, and common diseases that are influenced by both genetic and environmental factors, such as diabetes. The subtleties of biological processes involved in disease formation and progression, on the other hand, are still being researched and will soon be available when we have a big database of genomes to examine the relationships

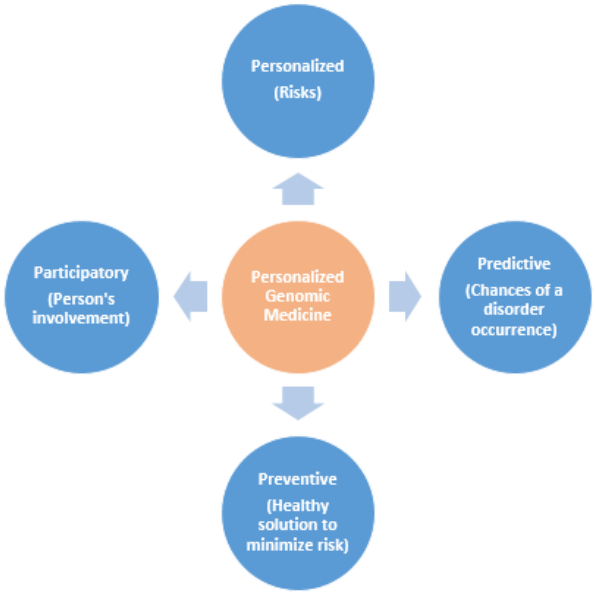

Fig 3. Salient features of Personalized genomic medicine

It has spawned pharmacogenomics [26] a branch of precision medicine that aims to tailor drug use for therapy [41]. Biomarkers were only mentioned in a few clinical trials conducted before 1990 [42], but this number has risen since 2005. Biomarkers are being identified and exploited in medication research and discovery, clinical trial design, and therapy in personalized medicine. Apart from pharmacogenomics (individual response to therapy), genomic science also gives information on genomic susceptibility (the likelihood of getting a disease). Personalized genomic medicine focuses on offering more specific insights into the body's functioning so that medical decisions can be made with more confidence. In [43], a comparison is drawn between reactive evidence-based therapy and proactive P4 medicine. The paper [7] outlines clinical concerns that may emerge during the therapy process, as well as the technologies that may be used to answer them and what decisions can be taken based on test results. The information gathered by these technologies aid in the development of targeted medicines. A pharmacogenomics test of HLA-B*1502 for epilepsy patients, for example, can tell if a patient is likely to suffer skin responses after taking carbamazepine.

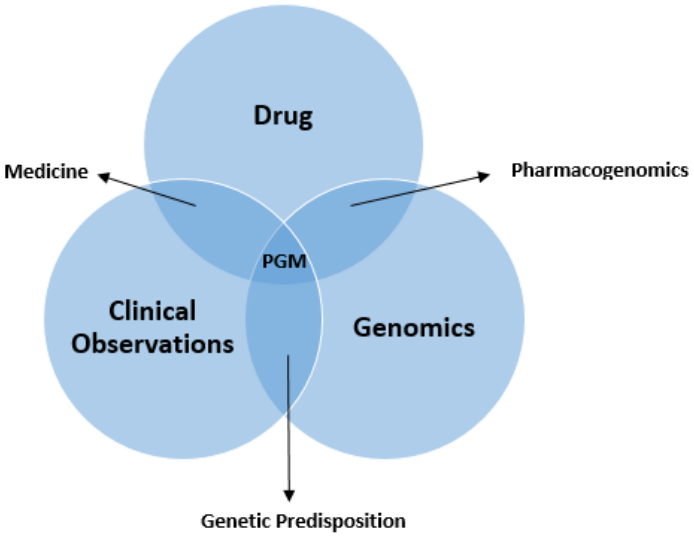

Fig 4. Personalized Genomic Medicine and its relationship with other fields

The main purpose of personalized genomic medicine is to:

Mitigate and control a medical condition: It aids in the development of a treatment plan that takes into consideration a person's unique genetic traits in addition to other clinical factors.

Reduce medicine side effects: Genomic data answers the question of what works and what doesn't for a patient with a specific medical condition.

Provide a P4 medical approach: This aids in finding the appropriate drug for the appropriate patient and delivering it at the appropriate time. It reduces the usage of inefficient and expensive medications, reducing the risk of unwanted side effects [19].

Avoid clinical trials: It helps to avoid clinical studies that will not work for a person with a specific genetic profile, as well as the mental stress that a person will experience throughout the study, as well as the wasted time and money.

Patient empowerment: It empowers patients by giving them with the knowledge they need to make informed decisions about treatment options and potential side effects.

Facilitates expert-mediated decision-making: Rather of relying solely on clinical test results, it can assist doctors in making more informed decisions.

However, there are misconceptions in people's mind regarding adoption of genomic medicine that hinders its utilization in clinical use. The following are some common misconceptions and fears about the use of genomic medicine in clinical decision-making:

- There is a fear that genomics would reveal everything about a person, even information that you don't want to share with others. However, whether they disclose it or not is entirely up to them; anonymity can be kept, just as it is in a typical clinical setting between a doctor and a patient.
- It can sometimes impede healthcare progress since genomic medicine is sometimes conflated with personalized medicine. However, genomic medicine is one of the tools of personalized medicine; otherwise, there are other non-genomic methods that provide evidence-based data that can be utilized to provide additional treatment.
- Patients' control over their health may be taken away by personalized genomic medicine, which may limit their treatment options. If a health insurance company chooses not include a prescription that hasn't worked well in the past for persons with a certain genetic composition, the knowledge gained from both genomic susceptibility information and pharmacogenomics information could have a detrimental impact on patient control. RonniSandroff, editorial director of Consumer Reports' Health and Family division, admitted this in a comment on the story [44]. However, such incidents can be avoided by enacting appropriate legislative laws, and the benefit of having complete awareness of a medical problem and the options available to a patient cannot be overlooked.
- Population classification as genetic superfamilies or based on racial or ethnic groupings may result from personalized genomic therapy. However, anonymity could be preserved to avoid discrimination. Genomic data is personal to an individual, and it cannot be shared without the patient's permission. This can be achieved by enforcing appropriate ethical and social rules.

Diagnostic firms, university, government, pharma and biotech corporations, and research labs are among the primary stakeholders in personalized medicine. The following are some of the significant developments that aided PGM:

- Projects like FP7 MyHealthAvatar [45] and DISCIPULUS [46] have demonstrated the value of digital clinical information in providing complete data for a patient, allowing for comprehensive insights and analysis of the patient's health.
- OpenEHR [47] is a standard providing an open-source approach for implementing an electronic health record (EHR) for clinical usage [48]. It can resolve interoperability issues establishing standards for sharing healthcare data.
- Projects such as IGNITE (Implementing Genomics in Practice) [49] and Vanderbilt PREDICT [50] focus on storing genetic data in EHR (Electronic health records) and providing clinical decision assistance.
- National Institutes of Health (NIH) initiated the All of US Research Program [51], [52] with the goal of collecting data from at least 1 million US volunteers. The enrolment for the programme commenced in the year 2018. The main goal is to collect data that may be utilized by researchers to study a wide range of ailments and enhance health services.
- The eMERGE (Electronic Medical Records and Genomics) network [53], and the Pharmacogenomics Research Network (PGRN) [54] both aim to catalyse precision medicine research by discovering genomic variation and applying it for therapeutic purposes, as well as minimizing pharmacological side effects
- The CLIPMERGE [55], 1200 Patients project [56] and PG4KDS [57] projects all focus on implementing pharmacogenomics models in clinical settings for prescribing personalized medicine.

- The ENCODE (Encyclopedia of DNA Elements) [26] project, , which began in 2003, focuses on studying the functional regions of DNA and making the data accessible on the ENCODE portal. ENCODE 2 and ENCODE 3 are two further phases that have been completed. This effort involved whole-genome studies of the human and mouse genomes.
- The P4 Medicine Institute is a non-profit corporation founded by Ohio State Medical School and the Institute for Systems Biology. It is made up of a network of clinical centres (about six) that collaborate with ISB to deploy P4 medicine strategies and tools [43]. Baylor College of Medicine, Cleveland Clinic Genomic Medicine Institute, and Personalized Medicine Alliance are among more academic medical institutions that support personalized medicine.
- Tailored Genomics initiatives are underway at several institutions in the United States [8], [58], with the goal of developing a model for implementing personalized medicine. Marshfield Clinic, St Jude Children's Research Hospital, the University of Maryland, University of Florida Health Shands Hospital, Mayo Clinic, University of Chicago, Indiana University, and others are just a few examples.
- An effort has been discussed in [59] for Europe for the digitisation of health data, bio-banking, screening, advanced imaging, next generation sequencing, biomarkers and analytics data to enable early diagnosis and enhance research and development.
- Some companies, such as Nebula Genomics, EncrypGen, and LunaDNA, have created a DNA data marketplace to allow customers and other institutions, such as pharma companies, to share genomic data [33], [60].

The majority of the efforts [8] regarding personalized genomics have been seen in developed countries currently and the resources needed for clinical sequencing, diagnosing rare disease, and identification of microbial pathogen is available in specialized centres only. The United States and Europe, which have a more developed framework, are concentrating on incorporating variant data into the electronic health record, discovering gene-drug pairs, and developing the clinical decision support component. They have also focussed on language standardization to be used for communication and regulations for data sharing. Developing countries have also begun projects in this direction, but due to a lack of resources and awareness of the technology, it will take longer for it to be fully functioning.

4.1 The Need of Personalized Genomic Medicine

The significance of personalized genomic medicine in healthcare can be understood through the following benefits that covers the scope of its usage also. The PGM can be used utilized for -

- Drug dosage Control for targeted therapies - A patient's drug dose may be determined by his genotype [13]. The FDA-approved targeted medicines for the treatment of various malignancies were reviewed in the study [14]. In [61], a case study of genotyping CYP450 genes to provide improved guidance in SSRI prescribing and dosing was presented.
- Genomic profiling- It has the potential to give genomic-guided healthcare information. Engineering techniques that assess both genetic and non-genetic aspects to deliver a better precision treatment were described in a review in [62]. This information can be utilized to determine the likelihood of cancer spread and tumour relapse [63].
- Companion diagnostic test – These tests are used to match a patient's symptoms to a certain medication. Medical records, including past drug histories, may aid in the avoidance of costly therapies (such as chemotherapies) and even the minimization of harmful procedures [64]. For example, Companion diagnostic assays (such as Ventana HER2 Dual ISH DNA Probe Cocktail) for Herceptin treatment and HER2 biomarker detection in breast cancer.
- Personalized lifestyle changes- Genomic data can be utilized to make healthy lifestyle changes and even to prevent medical conditions. It can aid in the identification of predictors of a person's reaction to the link between pharmacologic treatment and their lifestyle choices [63]. For example, Many research [65] have established the role of genes in Diabetes Mellitus and the risk factors linked with it. It can aid in detecting the link between genetic make-up and dietary changes, as well as their impact on the development of obesity, Diabetes Mellitus 2 (DM2), and metabolic syndrome.
- Disease prevention - It focuses on disease prevention based on a person's genetic information. If a person's genetic makeup predicts their risk of acquiring a disease, some diseases can be avoided by making changes in lifestyle.
- Lowers cost - It lowers the expense of clinical trials [66], [67] thus helping a person economically. It eliminates the need for trial-and-error treatment by matching patients to drugs. It aids in the unique diagnosis of several illnesses with similar symptoms.
- Drug efficacy - The efficacy of drugs can be improved, if complete genetic details of a person are available. It comprises pharmacodynamics (what a drug does to the body) and pharmacokinetics (what a body does to drug).
- Reduces drug trials - When compared to a non-personalized medicine strategy, it can minimize the overall cost of a drug usage process by reducing the number of unsuccessful drug trials, drug approval time, prescription length and frequency, and adverse drug reactions [66], [68].

- Identify varying responses to treatment - It can detect ethnicity-specific inter-individual variability in medication response. For example, before administering carbamazepine, all Asians should be tested for the HLA-B*1502 allele, which has been linked to illnesses such toxic epidermal necrolysis (TEN) and carbamazepine-induced Stevens-Johnson syndrome [69].
- Aid in drug development - It can assist the biopharmaceutical industry in developing genetically tolerant medications, reducing drug development costs and time [42].
- Identification - It can be used to identify a person in forensics. Since every person has unique DNA makeup that can help in identifying unknown samples or person.
- Genealogy- Genetics can also be used to identify biologically linked persons. It can be applied to the revelation of family ties.

4.2 Challenges of Personalized Genomic Medicine

The personalized genomic medicine is no doubt improving the quality of healthcare. However, for better utilization of genomic bog data and its translation in healthcare requires addressing of issues including, heterogenous data collection and standardization, data curation, de-identification & anonymization, consent management [9]. Major challenges that need to be dealt with for the adoption of personalized genomic concept in clinical system are summarized below.

Patient and Provider education- Due to the novelty of this technology, it is vital to educate patients about genetic testing, as well as healthcare professionals about genome-based medicine.

Healthcare Practitioner training- Well-trained practitioners with genomics and engineering training are required [62].

Sequencing Costs – The sequencing costs are constantly reducing, but they have a long way to go before they are affordable to everyone (now available at $10,000).

Data Privacy and Protection- The data protection [70] is important because it is personal to an individual and should not be easily accessible to others for use or misuse. Also, the data should not be changed in any way so that the results can be replicated and correlated with the new experiments.

Data Acquisition and Integration– To minimize problems linked to genetic data fragmentation, the data obtained from the sequencing facility must be shared unaltered with consumers and companies. Standardization is also required for the integration of data from various sources [14].

Data Ownership & Access Controls- Genomic data should not be shared without the authorization of the owner. The data must be completely under the control of the owners.

Genomic Big Data Storage- The genomic data is measured in gigabytes. The amount of data created is determined by the technology and its intended application. As a result, the storage reduction factor must be considered without causing any data loss. Apart from that, additional aspects of big data must be addressed before adopting a PGM system, such as the continually increasing volume of data generated by real-time analysis, the variety of data gathered and its interpretation [14], [71].

Human Ethics, Legal and Social Regulations [18], [43] – Data must be used in a way that is ethical. It should not be used to invade someone's privacy or cause harm to anyone. Before incorporating PGM into clinical practice, these difficulties must be addressed.

Data Analysis – Appropriate technologies should be created to allow for quick analysis of such large amounts of data while ensuring that no significant information is missed.

Quantitatively numerous Genetic variations- The handling of such a large number is difficult due to the difficulty of variant analysis and filtration in the whole genome [72].

Systemize and Structure the Genomic findings - PGM is currently only employed in a few clinical contexts. There is a need to systemize and structure the genomic discoveries, so that they may be shared and used for better medicine dosage and targeted therapy [73].

Development of Comprehensive Infrastructure and Technology- In order to integrate genetic testing into clinical practice, it is necessary to establish Comprehensive Infrastructure and Technology. It contains tools for health risk assessment (HRA), family health history (FHH), genome data management, and clinical decision support [18].

4.3 Application areas of Genomic information

This section discusses the application areas where genomic data can be utilized to improve the outcomes and further advancing health services.

- **Direct-to-Consumer Services:** The **Direct-to-Consumer (**DTC) movement [74], [75] offers services to consumers

who want to learn more about their bodies and their disease vulnerability. The use of genomic data to track kinship is becoming more common. It also provides a platform for data exchange as desired by the user. A variety of products are available that provide various amounts of information, such as genotyping particular variations, SNP chips, DNA sequencing, or whole genome sequencing (WGS).

- **Targeted Genotyping of Specific Variants:** Some companies are providing information on variants relevant to one or a few specific diseases or traits. For example, the company HairDX (Irvine, California(CA)) [74] provides information regarding genetic variants linked to hair loss.
- **Targeted therapies:** Therapeutics aimed at affecting specific biochemical elements or pathways that cause a disease or medical condition are known as targeted therapies [76]. Herceptin, GLEEVEC, Erbitux, Tamoxifen, and Avastin are some examples.
- **Companion diagnostics:** Companion diagnostics, also known as theragnostics, are used to establish whether a targeted pharmacological therapy is appropriate for an individual with a specific ailment. HER2 and Herceptin testing are two examples [77].
- **Pharmacogenomics:** Pharmacogenomics is the use of genomic information to predict or study an individual's reaction to a medicine. It lowers the chances of a patient's safety being jeopardized by a drug. CYP testing can be used to determine morphine sensitivity and warfarin metabolism, for example [78], [79].
- **Translational bioinformatics:** Translational bioinformatics is concerned with the development of methods to handle the storage, processing, and interpretation of biomedical data in order to meet P4 medicine goals. Both translational research and biomedical informatics (BMI) research [48] can help address the issues raised by the President's Council of Advisors on Science and Technology in the United States (PCAST).
- **Cancer risk and prevention, Diagnosis, Treatment:** Precision medicine can be used to determine the tests and treatments effective for specific malignancies. [57]. Precision medicine can assist doctors in preventing cancer, identifying individuals at high risk of cancer, diagnosing cancer early, selecting the best treatment option, and evaluating a treatment on a therapeutic basis [58], [80]. Breast cancer, colorectal cancer, lung cancer, esophageal cancer, some types of leukemia, certain types of lymphoma, ovarian cancer, stomach cancer, thyroid cancer, and others are among the malignancies for which precision medicine is being used to make treatment decisions.
- **Drug-Design:** Genomics can assist identify gene targets for a medical ailment and aid in the development of new drugs that can reach multiple targets.
- **Drug Labelling for Clinical use:** The Food and Drug administration (FDA) included a number of clinically useful Personalized Genomics variants on drug labels. Personalized genomics information is included on the labels of some of the FDA and European Medicines Agency (EMA) approved drugs. For example, the FDA has updated the carbamazepine label to include HLA- B*1502 biomarker information for toxic epidermal necrolysis (TEN) and Stevens-Johnson syndrome. Around 362 drug–biomarker pairs (DBPs) have been found from 261 prescriptions for drug labelling [15], [81].
- **Informatics:** With the advancement of personalized medicine, genomic data can be stored in electronic health records to give useful information about large numbers of patients, their drug history, and responses based on their genetic composition [81].
- **Drug repurposing:** When a drug's label is known to induce adverse responses in other genotypes, it can be altered or tagged with genotype information [82].

5. ENABLING TECHNOLOGIES FOR PERSONALIZED GENOMIC MEDICINE

Engineers have invented and are still developing technology to support the idea of personalized and precision medicine, which is becoming increasingly popular. The study [83] discusses enabling technologies ranging from wearables for monitoring body metabolism to drug design and administration based on artificial intelligence. Artificial intelligence, Genomic analysis/companion diagnostics, Genome-guided therapy, Microfluidics/companion diagnostics, Genome editing tools, RNA therapy, Nanomedicine, Wearables, Digital Health, Biomaterials, and Electronic health genotyping are some of the emerging technologies or platforms used in personalized and precision medicine. These technologies are being used in Personalized Medicine to deliver personalized care. It would be easier to successfully individualize therapy on a genetic and phenotypic basis with more data obtained using various technologies. Personalized medicine depends on integration of multi-omics with the enabling technologies to reshape the domain of personalized healthcare domain. Below are some of the significant technologies that are being used to promote personalized medicine.

Next Generation Sequencing

Cost-effective whole genome sequencing and targeted genetic tests are possible and affordable with advancement in NGS

technology and sequencing costs. NGS tools allows large-scale screening, diagnostics, and variant discovery. It can be utilized to detect genetic mutations, structural variants, polygenic risk factor and biomarkers [73], [84]. No doubt, NGS has revolutionized the genomics field, enhancing understanding of complex diseases and support targeted therapies for the patients [85]. This technology can be integrated with advanced algorithms and techniques like Edge-AI and Big data tools to efficiently manage sequencing data and further accelerate interpretation and analytics for precision care.

Artificial Intelligence

Artificial intelligence enables extraction of patterns, inferences and associations from the medical data collected. The generated insights assist in making sensible clinical decisions in a short period of time. Artificial intelligence (AI) as a tool for analysis combined with precision medicine will transform healthcare [86]. The translational research can further use the information generated from patient's data (clinical reports, genetic reports, and other data regarding environment and lifestyle) or electronic health record (EHR) to identify markers, both genetic and non-genetic, for a disease study. AI can extract relevant insights from data from a variety of sources and formats, allowing medical practitioners to make more informed decisions. Artificial intelligence (AI) can be applied in either a virtual or a cyber-physical environment.

Some of the applications for artificial intelligence include FREENOME (early cancer detection with AI), BUOY HEALTH (an intelligent symptom checker), ENLITIC (AI deep learning for advanced analytics), BERG HEALTH (treating rare diseases with AI), ZEBRA MEDICAL VISION (AI-powered radiology assistant), BIOXCEL THERAPEUTICS (biopharmaceutical development through AI), and others [87]. Artificial intelligence assistance may aid in the reduction of diagnostic errors as well as the extraction and maintenance of data. With the development of IBM's Watson and Google's Deep Learning [88], AI performance has improved. Additionally, AI-based diagnostic tools for diabetic retinopathy have received FDA approval.

The new emerging improvements [89] in tools such as pattern identification and natural language processing (NLP) will enhance capabilities of AI in drawing relevant insights and also it will be bias-free in reasoning and prediction. Quantum systems [90] that use custom machine learning algorithms will outperform conventional systems even more. The ultimate goal is to reduce illness burden and healthcare costs by preventing and early detecting diseases.

Machine learning

Machine learning, a different sort of AI [91], has been applied in healthcare for variety of applications, such as clinical decision support systems, personalised medicine, smart record keeping, medical imaging, behaviour adjustments, robotic surgery, and more . Microsoft's InnerEyeproject, for example, employs machine learning technologies to distinguish between healthy cells and malignancies. IBM's Watson oncology technology uses the patient's medical history to provide numerous therapy alternatives. Somatix aids preventative medicine by tracking a patient's everyday activities in order to enhance their lifestyle. Machine learning approaches are also being used to analyse data of 'multi-omics' type [92].

In resource-constrained environment, machine learning models can run directly on edge devices enabling real-time genomic and diagnostic analysis. , health monitoring and data sharing in real-time using wearable devices powered by edge-AI systems [93]. Edge-AI can be utilized for real-time genomic data processing and decision-make at the point-of-care, thus enabling personalized care and improving patient outcomes.

Big Data Platforms

Big data platforms supporting artificial intelligence and machine learning has made possible faster data processing and maintenance. For data extraction, data sharing, and data curation, there is a need to address the linked difficulties of data storage, computing or processing speed. When combined with deep phenotypic data [94], the obtained genomic big data organized and presented in a relevant and appealing way would aid in a better understanding of biological processes. Data provided by molecular profiling technologies (NGS, metabolomics, and proteomics) in particular can boost translational research to improve clinical care. Big data systems tools such as Apache Hadoop and Apache Spark allows scaling-up of storage and processing, empowering personalized medicine [95].

Cloud-based Genomic data platforms

Cloud-based genomic data platforms empower researchers to access and analyse data ubiquitously. Cloud computing coupled with advances in storage and computational power supports multi-omics big data management [39]. AWS Genomics, Google Cloud Life Sciences, Galaxy, Bionimbus, and DNAnexus are some cloud platforms [96], [97] for genomic data management, supporting collaborations and compliance with regulatory standards (HIPAA, GDPR). Cloud computing paradigms can further accelerate research enabling access to pre-configured pipelines and federated learning models in bioinformatics. These platforms pave the way for integration of genomic data into clinical workflows allowing for tailored treatment strategies. In addition, Efforts such as NIH-funded cloud platforms support biomedical data storage and analysis,

Data science

Physicians can utilize data science algorithms or techniques to uncover correlations between biomarkers, patient genetics, and treatment, similar to how data science is employed in many commerce applications to research people and purchasing patterns [98]. It has the potential to provide new insights on the biological processes involved in illness progression and prognosis. It can support remote patient health status monitoring and early disease symptom detection. Through various tactics, such as data mining algorithms, it can be easier to investigate numerous cases of a similar or dissimilar kind. It not only streamlines the healthcare workflow but also assists in promptly averting hazards and emergencies. In conclusion, data science can support technological improvements, shorter treatment times, lower treatment costs, and effective management of healthcare big data.

Bioinformatics

When DNA sequencing becomes even cheaper in the coming years, bioinformatics will be able to improve the therapeutic value of genomic data [98]. Bioinformatics can be used to analyse data from sequencing technologies or other sources in order to find gene variants or genetic changes that can be utilized to investigate the influence of a drug on patient prognosis [99]. It applies a variety of informatics tools to gain a molecular understanding of the medical problem. Using data science and AI-based methodologies, bioinformatics tools can further augment clinical decision-making.

Biosensors

The health of a patient can be remotely monitored using biosensors. The real-time data gathered can be utilized to provide tailored therapies. Aside from vital signs, biomarker data can be utilized to continuously track prognosis rate and send notifications if a certain threshold is achieved. One example is the ACCU-CHEK [100] commercial hand-held sensors, which require blood samples to monitor body's glucose level. The readouts from these instruments aid in the personalization of medicine dosage [83]. The digital biomarkers [101] or wearables meant for tracking a disease or monitoring bodily features such as heart rate readings, sweat analysis, blood pressure monitoring, glucose monitoring, and other physical characteristics have been developed [102], which can aid clinical trials.

Molecular diagnostics

Molecular diagnostics is the process of detecting and analyzing biomarkers for a certain health condition in order to better comprehend the disease mechanism [103]. Some of the most commonly used technologies for molecular diagnostics [104] includes fluorescence in situ hybridization (FISH), next-generation sequencing (NGS), microarrays, polymerase chain reaction (PCR)-based methods, nano-biotechnologies and mass spectrometry. These technologies help to further improve and provide a better treatment to the patient.

Blockchain Technology

ny application scenario that requires data to be saved as immutable, secure, and verifiable can benefit from Blockchain technology. Furthermore, data can be exchanged in a decentralized and accountable manner. Because of the way medical data or records must be preserved and shared with stakeholders, blockchain technology has recently found its use in the healthcare industry [105]. The blockchain technology has found many applications in healthcare such as secure data sharing, supply chain, data interoperability, data control, etc.

Digital Twin Technology

Digital twin models enable creation of patient's virtual replica at genetic level. These models can integrate genetic data with clinical data to a enable medical practitioners to handle a patient's treatment effectively, aid in refining treatment strategies, and a virtual testbed for precision interventions [106]. It can significantly reduce time and cost for adopting new treatment plans in market. Digital Twins can assist in identifying disease indicators, disease likelihood, and progression. However, ethical dilemmas need to be resolved before moving towards advanced health systems utilizing digital twins [107], [108].

Clinical Decision Support Systems

Clinical Decision Support Systems (CDSS) can leverage genomic data in integration with EHR data, medical databases, and advanced algorithms, utilizing data analytics for informed and evidence-based decision-making process [109]. These systems are capable of translating complex genomic data and draw useful insights to aid clinicians for better diagnosis, treatment, prognosis and prevention methods for the patients. CDSS in integration with genomics benefits in identifying potential drug-gene interactions, thus minimizing chances of adverse effects on patients during treatment. In short, CDSS enables early disease detection, personalized treatment recommendations, improved diagnosis, and enhanced drug selection. Watson for Oncology, for instance, utilizes NLP and ML approaches designed for cancer patients providing recommendations for personalized treatment [95].

Quantum Computing

A recent computing paradigm, Quantum Computing has the potential to resolve computational challenges in genomic data

processes and applications (sequence alignment, variant interpretation, complex trait mapping and molecular simulation ,protein folding prediction, messenger RNA codon optimizations, polypharmacological model optimization, therapeutic target identification). This technology has the potential to reshape precision medicine, accelerating genomic analysis and drug discovery [110]. However, there is a need to address technological barriers (hardware limitations, noise & decoherence, scalability, interoperability) to quantum computing to completely leverage it in medicine and healthcare [111].

6. DISCUSSION

The personalized genomics is no doubt has become a powerful tool for advanced healthcare system, in integration with big data analytics tools for data analysis and trustworthy platforms such as blockchain for data governance. Apart from its use in clinical genomics, the relevance of personalized genomics can be seen in various use-cases, for instance in pandemic situations like COVID 19, information collected through Genomic research can contribute in better understanding of the disease structure, symptoms, susceptibility, severity, prevention, control, and treatment. The biomarker or gene data can help in identifying a person's genetic susceptibility to disease infection and response to a treatment method, which provides aid in providing personalized treatment to both, infected- and likely to be infected individuals. Another use-case is DNA marketplace where commercial companies such as 23andMe, DNAtix, Nebula, etc. are providing various services such as genealogy and direct healthcare services to its consumers based on their DNA data.

All these efforts aim to provide a healthy and enhanced lifespan to the individuals. Genomics can enhance an individual's lifespan by identifying disease risk, especially in the cases of hereditary disease. Different diagnostic tests are available at different stages of life such as preconception, prenatal, newborn screening, paediatric medicine and adult/elderly medicine [112]. Apart from diagnosis, Genomics can be integrated with clinical practice for disease susceptibility, prognosis and therapeutic decisions, and monitoring [113]. However, there are still some challenges yet to overcome for clinical genomics such as technical (cost, result sensitivities) and interpretive barriers (variant interpretation) [112]. Another challenge is ethical concerns that needs more attention to increase trust in clinical genomics, which can be handled through proper rules and regulations for ensuring data privacy and sharing. One more major concern is clinical relevance of genomics that is whether genomic data is relevant for identifying risk of multi-genetic diseases, similar to that of monogenetic diseases. With multiple platforms and consumer services, the doubt on relevance of genomic data is due to lack of validity, reliability and utility [33]. However, these problems can be handled through proper standardization and involvement of medical professionals in the system [33]. To establish genomics as the core element of personalized healthcare, technologies including AI, Deep learning, blockchain, and other emergent technologies can be utilized to resolve technical challenges and regulatory challenges to enhance patient engagement, and improve personalised health services.[114]

7. CONCLUSION

To advance towards a future with a higher rate of illness prevention and disease-free living, personalized genomic medicine must be realized in order to live a healthy lifestyle supported by a person's genetics and an enhanced rate of cure if a disease or condition is detected early. In conjunction with big data, machine learning, and data science technologies, genomics can speed up the precision and personalized health care idea, which is still in development. Most drugs will be affected by this concept in the near future, when most of us will have our entire genomes sequenced and saved as medical data. This concept will require investment in three primary stakeholders: pharmacogenomics, molecular medicine, and health information technology. Appropriate practices and norms must be established so that everyone can participate in this system without doubt or stress. Of course, the focus of the healthcare system in the next decade will be 'omics'-based, with biomarkers, advanced algorithms based on artificial intelligence, and health-based apps. With various health monitoring technologies such as wearable devices and point of care devices/sensors feeding real-time data into these electronic health cards, e-genetic report cards will soon be a reality. As a result, addressing all of the issues associated with personalized genomics before the deadline becomes unavoidable.

8. ABBREVIATIONS

AI: Artificial Intelligence

BARD1: BRCA1-associated RING domain protein 1

BMI: Biomedical informatics

BRCA: Breast Cancer gene

BRIP1: BRCA1-interacting protein 1

CLIPMERGE: Clinical Implementation of Personalized Medicine through Electronic Health Records and Genomics

CNV: Copy number variations

CYP: Cytochrome P450

DNA: Deoxyribonucleic acid

DTC: Direct-to-Consumer

EBI: European Bioinformatics Institute

GWAS: Genome-wide association studies

HER: Electronic health record

HGP: Human genome project

HapMap: Haplotype Map

EMA: European Medicines Agency

ENCODE: Encyclopedia of DNA Elements

Emerge: Electronic Medical Records and Genomics

FDA: Food and Drug administration

FHH: Family health history

FISH: Fluorescence in situ hybridization

HER2: Human epidermal growth factor receptor 2

HLA-B: Human leukocyte antigen complex, class I, B

HRA: Health risk assessment

IBM: International Business Machines Corporation

IGNITE: Implementing Genomics In practice

ISB: Institute for Systems Biology

ISH: In situ hybridization

NGS: Next-generation sequencing

NIH: National Institutes of Health

NLP: Natural language processing

P4: “Personalized,” “Predictive,” “Preventive,” and “Participatory”

PCAST: President’s Council of Advisors on Science and Technology

PCR: Polymerase chain reaction

PET: Positron emission tomography

PGM: Personalized genomic medicine

PGRN: Pharmacogenomics Research Network

PMI: Precision Medicine Initiative

RNA: Ribonucleic acid

SNP: Single nucleotide polymorphism

SSRI: Selective serotonin reuptake inhibitors

TEN: Toxic epidermal necrolysis

US: United States

VUS: Variants of unknown significance

WGS: Whole genome sequencing

Statements and Declarations

Ethics Declaration

Not applicable

Consent for publication

Not applicable

Availability of data and material

Not applicable

Declaration of Competing Interest

The authors state that they have no known competing financial or personal interests that would have affected the quality of the work done in this study.

Authors' contribution statement

A. Arya: Conceptualization, Investigation, Methodology, Writing – original draft.

A. Malik: Conceptualization, Methodology, Writing – review & editing, Supervision.

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