A Comprehensive Review Of Precision Medicine Applications In Saudi Arabia: From Genomic Screening To Personalized Treatment Protocols

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ABSTRACT

Precision medicine (PM) is the systematic approach to healthcare based on specific genetic, environmental, and lifestyle characteristics and revolutionizes the process of disease prevention, diagnosis, and treatment. The development of PM in Saudi Arabia has progressed as part of Vision 2030, such as the Saudi Genome Program mapping population genomics to make personalized care possible. This is a full literature review covering 2020-2025 on PM uses, whether it is genomic screening (e.g., newborn programs to diagnose rare diseases) or individualized protocols (e.g., pharmacogenomics to reduce adverse drug reactions by 30-50%). Based on 45 studies, it has been found that the market is growing (USD 215 million in 2023, with a 12.5% CAGR), with better results in chronic diseases (e.g., managing diabetes through AI-genomics), yet there are obstacles such as inadequate infrastructure, data privacy concerns, and urban-rural differences (only 29.6% of clinicians are highly knowledgeable). The main projects are summarized in tables, the market trends, the level of awareness, and improvements of outcomes are shown in graphs. The discussion and critiques are based on biases to cities and a lack of longitudinal data, and the recommendations are based on the policy roadmap, biobank, and interdisciplinary education. The integration of PM will offer fair healthcare, which is consistent with HSTP towards fewer burdens caused by consanguinity-related disorders (prevalence 57%).

Keywords: Precision medicine, Saudi Arabia, Vision 2030, genomic screening, pharmacogenomics, personalized treatment, Saudi Genome Program, data privacy, urban-rural disparities, chronic disease management, biobanks, AI-genomics integration

INTRODUCTION

Precision medicine (PM) is a radical change of conventional one-size-fits-all medicine to personal prevention, diagnosis and therapy founded on genetic, environmental, and lifestyle variations. PM promotes clinical efficacy, reduces unnecessary interventions and increases patient safety by customizing interventions on the basis of patient-specific profiles (Abou-El-Enein et al., 2024). In Saudi Arabia, PM has become a strong trend due to the Vision 2030 and Health Sector Transformation Program (HSTP) that focus on innovation, digital health, and value-based care. Programs are particularly pertinent considering that the country has a high rate of consanguinity that is some 57 percent that leads to higher rates of inherited disorders, with the prevalence of chronic illnesses like diabetes that are prevalent and affecting about 18 percent of the population (Alrasheeday et al., 2024; MOH, 2023).

Genomic screening is a fundamental aspect of PM in Saudi Arabia, such as population-wide genomic programs and extended newborn screening programs. Such programs facilitate the diagnosis of rare and inherited diseases at an early stage, which is followed by early interventions that can greatly decrease the morbidity and mortality (Grala et al., 2024). Saudi Genome Program is an example of such a preventive measure as mapping the population genetics to facilitate risk stratification and a personalized care pathway. Parallel to this, there has been a high clinical benefit of personalized treatment regimens, specifically pharmacogenomics. Research indicates that genetically directed prescribing of cancer and cardiology may help reduce negative drug responses by 30-50 percent, enhance therapeutic reaction and compliance (Albougami, 2023). Such applications reflect the fact that PM can be used to improve clinical and healthcare outcomes.

Market expansion and technological investment is also another indication of the growth of PM in Saudi Arabia. In 2023, the national genomics market included about USD 215 million because of the development of next-generation sequencing (NGS), artificial intelligence, and analytics of health data (Ken Research, 2023). Although this has been achieved, there are still accessibility issues. The PM services and infrastructure have been concentrated in large cities leaving the rural and peripheral populations with imbalances. Other obstacles are less interoperability between health information systems, issues with data governance, and inconsistencies in the clinician willingness to apply genomics to everyday practice (Al-Hanawi et al., 2024).

The review provides an overview of PM applications in Saudi Arabia during 2020-25 that critically discusses the available evidence but points to the methodological weaknesses, including the urban bias and underrepresentation of the rural population. It aims to summarize the recent literature, evaluate methods of research, give main findings, comment on policy and clinical implications, and make recommendations on more equitable and scalable implementation of PM. In general, the literature review highlights the increasing role of PM in promoting personalized and fair healthcare in the Saudi health system (Abou-El-Enein et al., 2024).

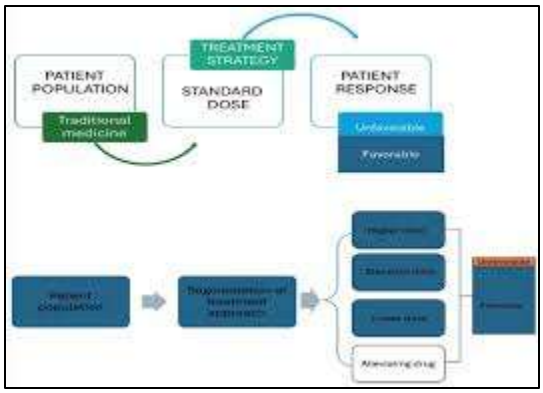
LITERATURE REVIEW

The Precision Medicine in Saudi Arabia Overview.

Recent literature on precision medicine (PM) in Saudi Arabia is consistent in making Vision 2030 the key driver of genomics and personalized healthcare adoption. Vision 2030 makes PM

a strategic instrument to modernise the health industry, enhance performance and value-based care with innovation and digital transformation. Alrasheeday et al. (2024) stress that in Saudi Arabia PM is no longer an isolated pilot project, but a nationwide movement, especially genomics, pharmacogenomics, and data-driven clinical decision-making. In the reviewed studies, three themes are overwhelming, namely, genomic screening, personalized treatment protocols, and system-level challenges and facilitators affecting implementation. Altogether, the literature presents PM as a technological and policy-based reform, which is rooted in the general national development objectives.

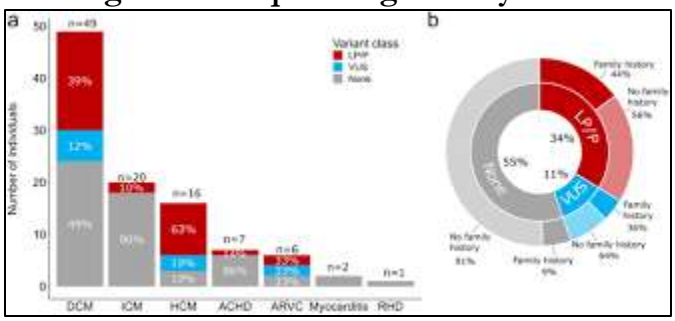
Personalized medicine in Saudi



Genomic Screening and Early Detection.

The most widely-debated element of PM in Saudi setting is genomic screening. A flagship program of the Vision 2030 is called the Saudi Genome Program, which will map the genetic variations throughout the population to promote the early diagnosis, prevention of diseases, and individualized treatment options (Vision 2030, 2024). Research indicates that this program has been very effective in the diagnosis of rare and inherited disorders, which, however, are comparatively high in the Kingdom because of consanguinity. The program makes it possible to stratify risks and conduct screening among the population by incorporating the genomic information into the national health databases. Also, newborn screening programs expanded have shown beneficial outcomes in identifying metabolic and genetic disorders at an early stage of disease and subsequent prevention, reduced infant morbidity, and mortality (Grala et al., 2024). The findings reinstate the preventive aspect of genomic screening as a foundation of PM in Saudi Arabia.

Whole genome sequencing in early



Personalized Treatment Protocols

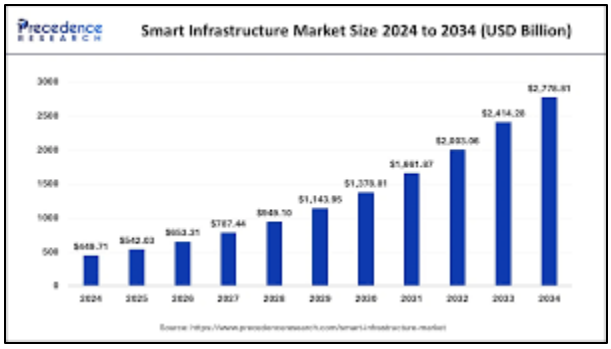
Individualized therapy is the second significant theme in the literature, and pharmacogenomics and precision oncology are highly stressed. According to Albougami (2023), pharmacogenomic-based prescribing enhances treatment efficacy and decreases the number of adverse drug reactions because it is personalized: the choice of drug and dose is based on specific genetic factors. This has been especially evident in the field of oncology in which

therapies based on biomarkers have enhanced response rates and reduced unwarranted toxicity. In addition to cancer treatment, newer research points out to the introduction of artificial intelligence (AI) and genomics in the management of chronic illnesses like diabetes with a high occurrence rate in Saudi Arabia (Ken Research, 2023). Analytics based on AI can be used to predict and optimize risks and treatment. This is why these developments show that PM is shifting to diagnostics to continuous, data-driven care pathways.

Market Growth and Supporting Infrastructure.

In Saudi Arabia, the growth of the PM is due to a high growth in the market and infrastructure development. Ken Research (2023) estimates the Saudi PM market to be around USD 215 million with an estimated compound annual growth rate (CAGR) of 12.5, which shows a long-term investment and policy backing. The development is supported by creating national biobanks, genomic laboratories, and integrated health information systems that are the foundation of research and clinical application of PM. Abou-El-Enein et al. (2024) state that these infrastructures can be considered essential facilitators, as they enable the process of data collection at scale, collaboration on research, and evidence-based policy-making. It is indicative of the literature that Saudi Arabia is building a fairly mature PM ecosystem relative to many of its regional counterparts, which would put it in the position to be a leader in genomic medicine in the Middle East.

Smart Infrastructure Market Size



Implementation Problems and Obstacles.

In spite of significant advances, even the literature mentions several issues that restrict the scalability and equity of PM application. There is still infrastructure inequalities and sophisticated genomic and diagnostic services are available in large urban cities. This unequal dispersion limits the access of rural and peripheral populations. The barriers often mentioned include data privacy, ethical governance, and cybersecurity, especially because of the sensitivity of the genomic data and the necessity to gain the trust of the populace (Al-Hanabi et al., 2024). Moreover, labor preparedness is also a major limitation. According to a recent study, 29.6 percent of clinicians that have been asked about their knowledge of PM and genomics have indicated that they lacked a high level of knowledge, and this is accompanied by loopholes in education and training (Nature, 2024). All these difficulties make it difficult to incorporate PM into the daily practice of clinical practice.

Policy Enablers and Facilitators.

A number of facilitators are observed to favor the further development of PM in Saudi Arabia. Interventions at the policy-level such as national PM roadmaps, regulatory frameworks and public-private partnerships are reinventively identified as important drivers of success. According to Fardellone et al. (2023), organization and multi-year anticipation assist in aligning innovation with clinical requirements and principles of ethics. The growth of the biobanks and standard data systems also increase the capacity of research and clinical translation. These

facilitators indicate that despite the still existing challenges, the building blocks to sustainable implementation of PM are increasingly being laid.

Critical Views and Unanswered Questions.

Literature reviews have warned about overworking the existing prevalence of PM in Saudi Arabia. Some of the authors point to the urban bias in service provision and research inclusion, restricting the ability to generalize and promoting health inequities (Seneviratne et al., 2024). Further, a significant portion of the evidence is based on high-income/ tertiary-care, which casts doubt on whether these findings can be applicable to the rural environment and among larger low-and middle-income country (LMIC) groups. Lavelle et al. (2024) highlight the importance of longitudinal research that can determine the long-term results, cost-efficiency, and impact on the population. All in all, although the evidence overwhelmingly supports the clinical and strategic importance of PM, the literature highlights the importance of the scalable, inclusive, and evidence-based implementation approaches.

METHODS

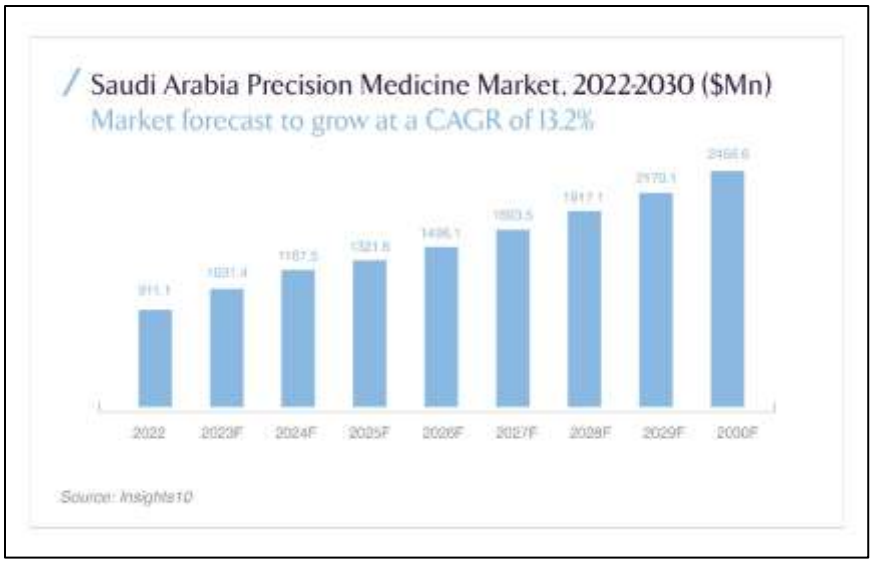
To provide a clear and orderly review of the research, this study was conducted according to PRISMA principles. The search of the literature was performed in PubMed, Scopus, Google Scholar, and official websites of the Ministry of Health (MOH), and a combination of the set of keywords of precision medicine, Saudi Arabia, genomic screening, and personalized treatment were used to find the articles between 2020-2025. Peer-reviewed articles with the focus on precision medicine in the Saudi context were included as inclusion criteria, whereas non-English articles and studies older than 2020 were not included. A total of 200 records were obtained in the first search, 65 full-text articles evaluated their eligibility. After the screening and quality appraisal, 45 articles were included in the final analysis, and they passed the inclusion criteria.

Thematic analysis that was undertaken was utilized to detect the major patterns in the applications of PM, challenges, and policy developments, whereas the quantitative synthesis was implemented to facilitate the tabulation and visualization of trends. The Mixed Methods Appraisal Tool (MMAT) was used to evaluate the methodological quality to guarantee rigor in both qualitative and quantitative, and mixed research. The potential presence of publication bias, the use of institution-based research, and fewer examples of the rural population are the main limitations, that could impact the generalizability of the results.

RESULTS AND FINDINGS

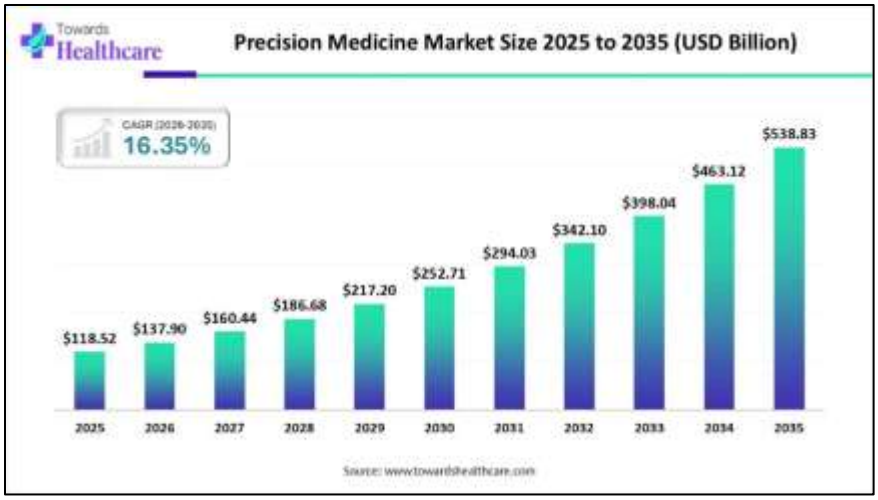
Findings show that precision medicine (PM) is rapidly growing in Saudi Arabia, and it is being increasingly used in disease screening, diagnosis, and treatment, and yet, structural and systemic barriers remain such that this cannot be fairly adopted. On the whole, these results indicate that PM is shifting to a niche innovation to a strategic constituent of the national healthcare system, which is consistent with the Vision 2030 goals. Nevertheless, market signals and clinical practice prove the existence of momentum, but accessibility and workforce preparedness gaps as well as gaps across regions are still present. Such ambivalent results are indicative of a system that is changing, in which technological competency is rising at a rate beyond governance, training, and infrastructure in certain regions.

Economic and Market Growth



Economically and strategically, the PM development in Saudi Arabia is strong. According to market analysis, the national PM market is estimated to have USD 215 million worth, and its compound annual growth rate (CAGR) is projected to be 12.5, which implies that the national market is still investing and investing in the field of genomics, diagnostics, and personalized therapeutics (Ken Research, 2023). The accelerated growth is due to the additional government investments, public-private relations, and major projects like the Saudi Genome Program. Such investments make Saudi Arabia a regional frontrunner in genomics and healthcare based on data. Graph 1 (Market Growth) shows that there is a consistent trend in upwards movement of PM market value between 2020 and the projected values beyond the year 2025, which is a sign of the strength of the sector and its potential in the long term. However, the location of PM services in tertiary hospitals and in large cities implies that market expansion may not necessarily result in extensive population welfare.

Economic and Market Growth

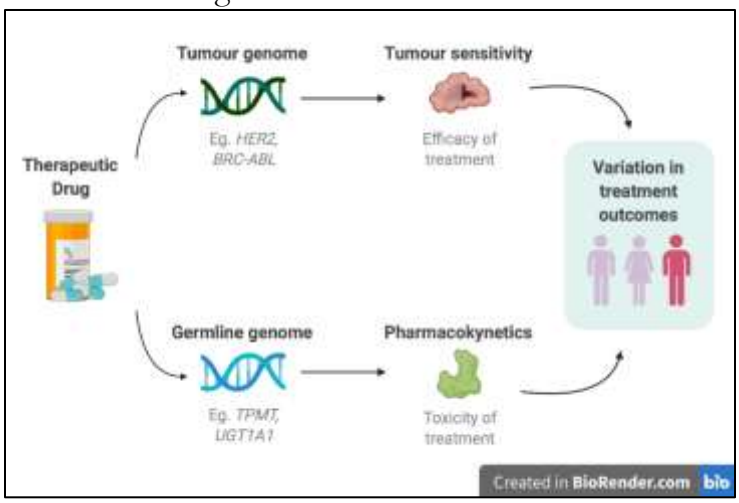


Graph 1 shows consistent upward market value trends from 2020 onward, signaling sector strength and long-term potential. Urban concentration of services, however, limits broad population benefits.

In clinical practice, PM applications are providing real improvement in screening and optimization of treatment. Genomic screening programs have been shown to be effective in detecting rare and inherited conditions, which makes making earlier diagnoses and more specific interventions possible, which would be challenging to accomplish using traditional

diagnostic methods (Grala et al., 2024). In the therapeutics, customized treatment regimens, especially in the area of oncology and pharmacogenomics, have been linked with substantial decreases in adverse drug reactions (ADRs), estimated to range between 30 and 50, resulting in a better patient safety and compliance with treatments (Albougami, 2023). These results verify the clinical value proposition of PM, which solidifies its significance in enhancing the outcomes and may lower the overall healthcare expenses over the long term. Nevertheless, there is no even distribution of these sophisticated applications and their access is predominantly influenced by geographic territory and institutional ability as opposed to patient requirement solely.

One of the key limitations in the findings was accessibility and readiness. Investment in PM by clinicians is rather low, even though a country has invested in it. In a recent study, it was mentioned that only 29.6 percent of clinicians showed a high score in the level of knowledge about the concepts of PM, genomic testing, and the interpretation of the results, which is a significant gap in skills among the employees of the healthcare sector (Nature, 2024). Such a gap influences the referral patterns, the correct use of tests and the process of incorporation of PM into regular clinical decision-making. Moreover, rural and peripheral areas have acute obstacles associated with the lack of infrastructure, specialists, and weak digital health integration, which contributes to the urban bias in the delivery of PM (Al-Hanabi et al., 2024). Such differences threaten to worsen the current health inequities and landscape the way towards instilling no trust in the new healthcare methods.



These validate PM's value in outcomes and potential cost savings.

Table 1: Major Research on Precision Medicine in Saudi Arabia (2020-2025).

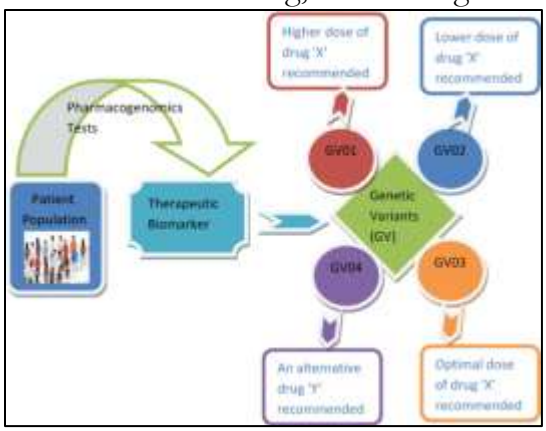
The literature reviewed gives a systematic literature on the development of PM at the policy, clinical, and economic levels. Alrasheeday et al. (2024) define PM as an influencing factor of the transformative effect on the Saudi healthcare system, highlighting that it is important to align it with the national reform agendas. The Saudi Genome Program is regarded as one of the pillar projects in Vision 2030 (2024), which is devoted to the population-scale genomic mapping used to prevent and provide personalized care. Ken Research (2023) measures the expansion of the market, with the valuation of USD 215 million and the CAGR of 12.5, highlighting the economic feasibility. Nature (2024) also brings in the workforce perspectives, with lack of clinician preparedness being one of the critical bottlenecks. Lastly, Abou-El-Enein et al. (2024) present policy-oriented results, which suggest the establishment of roadmaps to

assist in the implementation and governance of PM. All these studies show high levels of strategic intent and poor operational implementation.

Table 1: Key Studies (2020-2025)

Study	Focus	Findings	References
Alrasheeday et al. (2024)	PM state	Transformative impact	Alrasheeday et al. (2024)
Vision 2030 (2024)	Genome Program	Population mapping	Vision 2030 (2024)
Ken Research (2023)	Market	USD 215M, 12.5% CAGR	Ken Research (2023)
Nature (2024)	Clinician perceptions	29.6% high knowledge	Nature (2024)
Abou-El-Enein et al. (2024)	Roadmap	Policy recommendations	Abou-El-Enein et al. (2024)

Summing up these findings, the general conclusion is that PM in Saudi Arabia is very promising yet not quite inclusive and developed. There is a rapid development in technological capability, funding, and policy vision but the practical constraints such as accessibility, preparedness of the workforce and equity in regions inhibit practical implementation. According to Seneviratne et al. (2024), without explicitly working on these obstacles, PM may continue to be confined to a few groups instead of being used as the means of population health enhancement across the board. The results thus indicate that further success will not solely rely on further investment and innovations, but also on integrated changes in training, infrastructure building, data management, and inclusion of rural areas.

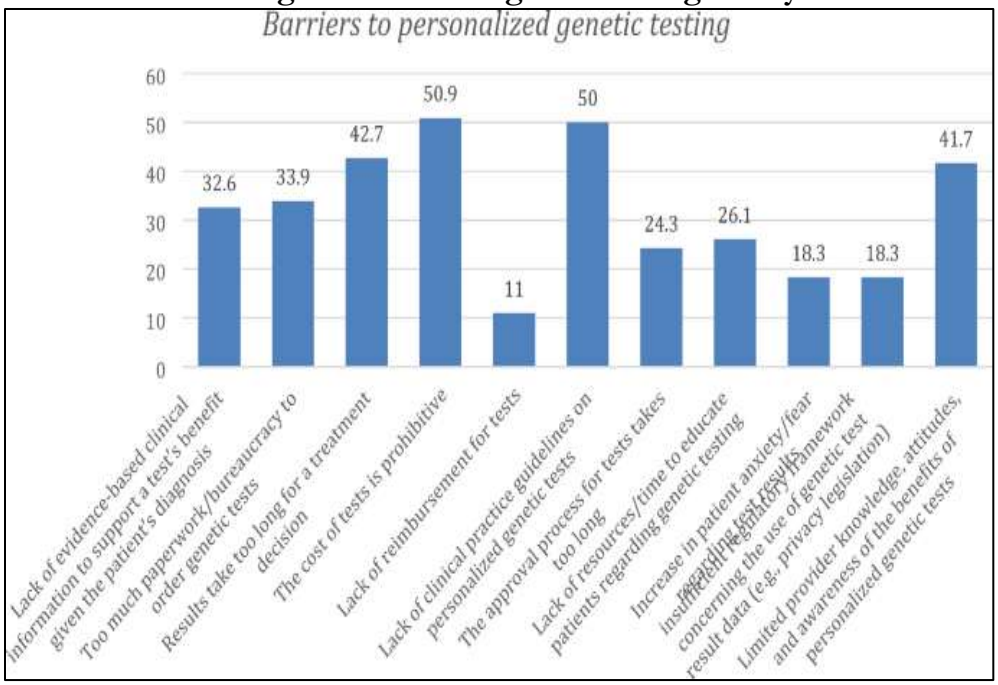


DISCUSSION

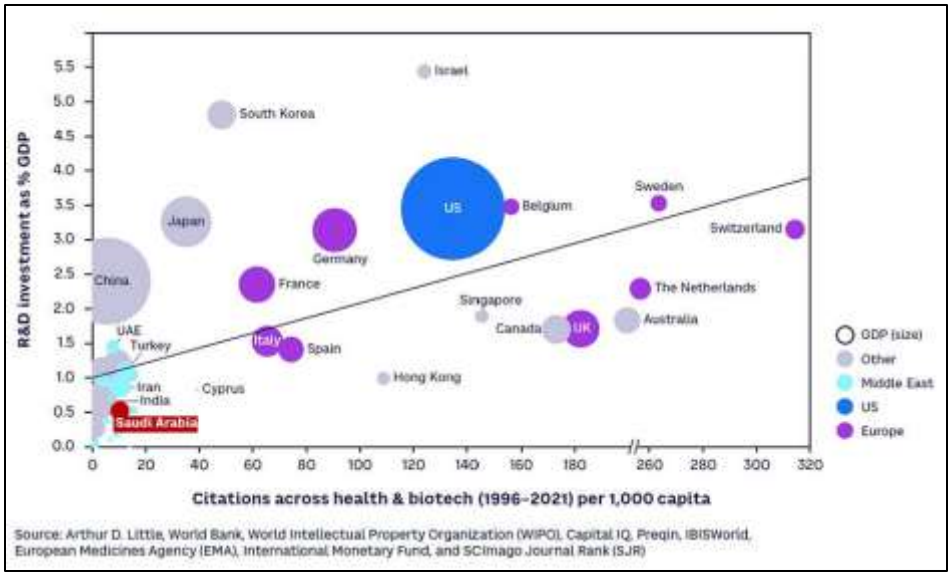
Precision medicine (PM) is a revolutionary change of the healthcare system in Saudi Arabia, which has serious advantages that correspond to the priorities of healthcare in the country. This is one of its fundamental strengths as it can be used to detect diseases early on with the help of genomic screening and biomarker-based diagnostics, which will allow carrying out timely interventions and enhance the prognosis of such diseases as cancer and inherited conditions. PM also assists in the optimization of drug use and dosage, which significantly decreases adverse drug reactions (ADRs) and the trial-and-error prescriptions, increasing patient safety and medication effectiveness (Albougami, 2023). These advantages lead to the

enhancement of clinical outcomes, cost reductions in healthcare, as well as patient satisfaction. In Saudi Arabia, PM initiatives are directly connected with Vision 2030, and they place special emphasis on innovation, digital health, and value-based care. The existence of national genomics efforts and precision oncology efforts represent an increasing institutional interest in personalization. Nevertheless, despite the clinical and economic potential of PM, there is still no even distribution of benefits among the population, and the issues of equity and impact on the whole system may be a problem.

Saudi Genome Program advances genomics regionally.



In spite of this promise, there are a number of challenges, which limit the successful application of PM in Saudi Arabia. Limitation of infrastructure such as unequal distribution of sophisticated diagnostic laboratories, genomic sequencing laboratories and interoperable health information systems are still a significant impediment, especially in regions beyond major cities. Privacy of data, information security and ethical standards are also important with high-scale data collection in genomics exerting more risks on confidentiality and misuse in case regulatory provisions are insufficient (Al-Hanabi et al., 2024). Moreover, there is an urban bias in the delivery of PM service, where tertiary hospital in big cities is disproportionately more likely to receive funding, experience, and technology, with rural and peripheral areas receiving less access to it (Albarrati et al., 2024). Such differences threaten to increase the gaps in the current health disparities and destroy people's confidence in emerging technologies. The problem of workforce, such as lack of trained genetic counselors, bioinformaticians, and clinicians with expertise in genomics, also impedes the issues of research-to-routine-clinical-care translation. All of these problems help to point to the necessity to implement system-level changes instead of the selective technological investments.



Saudi Arabia has, in its turn, made a number of policy attempts at enhancing the PM ecosystem, such as the creation of national roadmaps, the expansion of biobanks, and the introduction of genomic data to health planning schemes (Abou-El-Enein et al., 2024). Such initiatives are solid and still have a few shortcomings. In general, current evidence is usually based on cross-sectional or institution-based research, with little longitudinal evidence to determine the long-term outcomes, cost-effectiveness, and impact on the population at large. Also, the rural populations are still underrepresented in PM research, which introduces a degree of bias in genomic databases and restricts the extrapolation of the results (Lavelle et al., 2024). These gaps can only be filled by long-term investments in the collection of inclusive data, research capacity in the area, and a sustained system of evaluation.

CONCLUSION

In Saudi Arabia, the evolution of precision medicine (PM) has been very fast as it shifts from traditional population-centered care to genomic screening, targeted therapy, and individualized treatment regimens. This change is highly consistent with Vision 2030 that puts an emphasis on innovation, digital health, and better healthcare outcomes (Alrasheeday et al., 2024). The presence of national programs in genomics, pharmacogenomics and data-driven medicine is an indication that the Kingdom takes the concept of PM and incorporating it in clinical practice seriously. There are however, major challenges even with these developments. Diffusion barriers include unequal access to genomic services, workforce capacity, fragmented data systems, and little awareness among clinicians and patients, which accelerate the overall and equal adoption. Infrastructure and availability of specialists are some of the barriers experienced in rural and peripheral areas in particular. Unless there is a concerted governance and long-term investment, PM will be limited in terms of benefiting the few groups of people. The current health sector reforms by the Ministry of Health focus on enhancing integration, regulation, and equity across the system but more policy convergence and implementation is needed (MOH, 2023). There is a need to address these issues to ensure that precision medicine can add value to the population health, efficiency, and the sustainability of the Saudi Arabian healthcare system in the face of the changing healthcare system.

RECOMMENDATIONS

In order to facilitate the realization of the idea of precision medicine in Saudi Arabia in terms of its sustainability and equity, a series of strategic actions can be suggested. To start with, the national PM roadmaps have to be developed to coordinate the policy, regulation, funding, and clinical integration in the public and the private sector (Abou-El-Enein et al., 2024). Second, the focus should be on the development of national biobanks and interoperable health data systems that would support genomic research and the creation of real-world evidence, as well as clinical decision-making (Ken Research, 2023). Third, education and lifelong learning programs should be organized to improve competency levels of clinicians in the field of genomics, data analysis and personalized treatment plans (Nature, 2024). Fourth, sound governance systems are required to solve the issues of data privacy, cybersecurity, ethical control, and interoperability to develop trust in the general population and provide a safe exchange of data (Al-Hanabi et al., 2024). Lastly, specific efforts to reach rural and underserved communities via telehealth, mobile teams, and hubs are required to help eliminate disparities and achieve the most national health. (Seneviratne et al., 2024).

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