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The Role of Genetic Testing in Preventing Cardiovascular Diseases

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ABSTRACT

Cardiovascular diseases (CVDs) are still the most common cause of death globally, and early prevention and detection are essential to limiting their effects. The purpose of this research was to assess the potential of genetic testing in the prevention of cardiovascular diseases in patients at the District Headquarters Hospital Lodhran. 100 patients with CVD risk factors were enrolled and received thorough clinical evaluation, biochemical profiling, and panelized genetic testing targeting the most relevant genes for cardiovascular risk, such as LDLR, APOB, PCSK9, MYH7, MYBPC3, and CYP2C19. Analysis identified that 32% of the patients had pathogenic or likely pathogenic variants that are associated with higher cardiovascular risk. The most prevalent mutations were in LDLR and APOB genes, linked to familial hypercholesterolemia, and CYP2C19 polymorphisms that impact the efficacy of antiplatelet therapy. Those with genetic mutations had significantly elevated levels of LDL cholesterol and family histories of premature cardiovascular events. Genetic testing informed individualized interventions including maximized lipid-lowering treatment and individualized antiplatelet regimens. The report emphasizes the practicability and clinical value of introducing genetic testing in a district hospital environment within Pakistan, in spite of limited resources and poor public awareness. Integrating genetic screening into standard cardiovascular practice can facilitate early diagnosis, risk stratification, and prevention of disease, reducing overall burden of disease. Wider access to genetic services and education of patients are key measures to advance cardiovascular outcome in the region.

Introduction

Cardiovascular diseases (CVDs) are the greatest cause of morbidity and mortality across the globe, with millions of lives lost every year. The World Health Organization (WHO) reports that as many as 17.9 million individuals lose their lives every year to CVDs, representing 32% of the entire global deaths [1]. In Pakistan, cardiovascular disease burden has been increasing consistently because of changes in lifestyle, unhealthy dietary practices, urban living, and limited awareness of prevention strategies. Prevention and detection of CVDs have historically been based on evaluating clinical signs, lifestyle components, and general blood work[2]. But with the advent of genetic testing, a new aspect has been added to preventive cardiology so that earlier detection and targeted risk prediction according to a person's genetic profile is possible. Genetic testing is the process of examining DNA, the basic hereditary material, to detect mutations or variations which might lead to a tendency of developing specific diseases in a person. In cardiovascular health, such tests can identify inherited disorders like familial hypercholesterolemia, hypertrophic cardiomyopathy, arrhythmogenic right ventricular cardiomyopathy, and other gene mutations that are at increased risk of myocardial infarction, stroke, and other cardiac events. These findings enable both clinicians and patients to adopt preventive measures even before the development of symptoms[3].

The quick evolution of molecular biology, genomics, and bioinformatics has made genetic testing cheaper and readily available. What was once considered a tool at the exclusive domain of academic institutions or specialist medical facilities is now being implemented at clinical institutions, even district-level hospitals like the District Headquarters Hospital in the Lodhran district where this study was undertaken[4]. Lodhran, as a semi-urban region of South Punjab, has major challenges regarding healthcare infrastructure, availability of diagnostic facilities, and awareness among the population. Thus, carrying out this study in such an environment offered a chance to evaluate the practical application and viability of incorporating genetic testing into standard cardiovascular risk assessment in resource-limited settings[5].

The contribution of genetics to cardiovascular disease is complex. Some inherit single-gene mutations leading to monogenic disorders, e.g., familial hypercholesterolemia, resulting in high levels of LDL cholesterol from a young age. Others may inherit several genetic variants with a small contribution each, but their combined effect is to increase susceptibility—a polygenic risk**[6]**. Genetic testing, either by means of targeted gene panels or whole-genome sequencing, aids in the identification of both forms of risk, thereby providing a complete picture of an individual's susceptibility to heart disease. In practice, the identification of individuals at risk by genetic screening can result in early lifestyle interventions, pharmacologic therapy (e.g., statins or antihypertensive medication), and ongoing monitoring[7]. For instance, if an individual is diagnosed with a pathogenic mutation that predisposes to cardiomyopathy, routine echocardiograms and ECGs can be performed in order to track the heart function and catch early alterations. Furthermore, family screening becomes achievable, where blood relatives of the patient can also be screened and made aware of their possible risk. This cascade testing strategy is one of the most effective strategies in community health, facilitating the prevention of harm on a larger scale [8].

Another important benefit of genetic testing is its application to personalized medicine. Not every patient reacts in the same way to medication because of genetic differences in drug metabolism, effectiveness, and side effect reactions. Pharmacogenomics—the examination of how genes influence an individual's response to medication—has allowed doctors to personalize treatment plans according to one's genetic profile[9]. For example, in treating CVD, medications such as

clopidogrel, warfarin, and statins have an unpredictable response according to polymorphisms in genes including CYP2C19, VKORC1, and SLCO1B1. Through genetic testing, poor responders and potential adverse reactors can be detected, thus enhancing the therapeutic benefits and avoiding harm. In spite of the promise of genetic testing, its use in clinical practice, particularly in resource-poor nations such as Pakistan, is beset with problems. These are limited access to testing laboratories, expense, unavailability of genetic counselors, ethical issues, and social stigma[9]. Further, the medical infrastructure in rural and semi-urban regions usually does not have the facilities to accommodate sophisticated diagnostics. Pilot studies and research initiatives, like the study carried out at DHQ Hospital Lodhran, seek to fill this void by assessing the viability of introducing genetic testing in such environments. The study also points towards the patients' and healthcare professionals' awareness and acceptance of genetic testing. One of the key elements of preventive medicine is patient education and consent. Individuals need to comprehend the meaning of genetic results—not just for themselves but also for their families [10]. Likewise, doctors need to be well-versed to analyze genetic reports and incorporate them into clinical decision-making. We saw diverse levels of awareness in our study, and some effort from the research process went into informing participants and stakeholders about the strengths and weaknesses of genetic screening[11]. The ethical aspects of genetic testing cannot be overlooked. Concerns regarding informed consent, privacy of data, psychologic effect of test results, and the risk of genetic discrimination must be addressed with caution. Guideline development and standard operating procedures at the institutional and national levels are critical for the ethical incorporation of genetic testing into preventive cardiology[12].

Genetic screening has huge potential in the prevention of cardiovascular disease by allowing early identification of vulnerable patients and enabling targeted treatment planning. Admixture of genetic screening with standard clinical practice, particularly in district general hospitals, is a step toward the modernization of Pakistan's healthcare system. By conducting this study at the District Headquarter Hospital, Lodhran, we sought to assess the clinical, practical, and social dimensions of genetic testing among the local population. The results of this study not only add to the scientific knowledge regarding genetic risk factors in CVDs but also offer a model for the implementation of preventive genomics in public health systems in similar socio-economic environments.

Methodology

Study Design

This research was conducted as a descriptive cross-sectional observational study. The purpose of the research was to assess the utility of genetic testing in the early identification and prevention of cardiovascular diseases. The study examined genetic markers for cardiovascular risk and identified how this testing could be feasibly incorporated into daily cardiology practice within the setting of a district-level hospital.

Study Setting

The study was undertaken at the District Headquarters (DHQ) Hospital, Lodhran. Being based in South Punjab, the hospital caters to both rural and semi-rural patients and was selected because of the heterogeneity of its patients and representative nature of its population. The cardiology and internal medicine departments of the hospital were the main venues for data collection and patient interaction.

Study Duration

The research lasted six months, commencing from [insert start month and year] and ending in [insert end month and year]. This provided enough time to recruit, screen, test, and counsel participants in an organized and ethical fashion.

Target Population

The target population included adult patients visiting the outpatient departments of cardiology and internal medicine at DHQ Hospital Lodhran. The primary focus was on patients with a documented history of cardiovascular diseases or patients with a strong family history of cardiac conditions, as they were likely to be benefitted by genetic screening and preventive care.

Inclusion Criteria

Adults aged between 18 and 65 years were the participants in the study. Both males and females were included, as long as they had a diagnosis of a cardiovascular disease like hypertension, coronary artery disease, myocardial infarction, or arrhythmias. Participants with a family history of premature cardiovascular disease were also included. All the participants gave written informed consent and showed a willingness to be tested and counseled genetically.

Exclusion Criteria

Patients with terminal illnesses or other serious comorbid conditions that may affect cardiovascular risk were excluded. Pregnant women and those who refused to undergo genetic testing were also excluded. Patients who had already undergone genetic testing for monogenic cardiovascular diseases were also excluded to maintain consistency in the testing process and interpretation.

Sample Size

The sample comprised 100 participants who were recruited through non-probability purposive sampling. The sample size was deemed practical for a single-center trial carried out in a public-sector hospital within six months. It was also practical to enable significant statistical analysis and interpretation despite time and resource limitations.

Data Collection Tools and Techniques

Information was gathered using guided face-to-face interviews with the help of a pre-validated questionnaire. The questionnaire elicited demographic data, lifestyle practices, clinical history, cardiovascular disease family history, and the degree of awareness about genetic testing. The questionnaire was complemented by the completion of a basic clinical check-up, comprising measurement of blood pressure, body mass index (BMI), fasting blood glucose, and lipid profile. Blood was drawn in sterile EDTA tubes and samples collected for genetic analysis. The samples were subsequently taken to a certified molecular diagnostic laboratory for DNA analysis and extraction.

Genetic testing comprised screening for recognized genetic variants that confer risk for cardiovascular disease. This involved mutations in genes like LDLR, APOB, and PCSK9 for familial hypercholesterolemia, MYH7 and MYBPC3 for cardiomyopathies, and CYP2C19 for drug metabolism in the context of cardiovascular pharmacotherapy. Depending on the test and gene panel, methods like Polymerase Chain Reaction (PCR) and Next-Generation Sequencing (NGS) were employed for mutation detection.

Data on Preventive Outcomes

Genetic testing results led to appropriate counseling of participants based on those outcomes. High-risk individuals were counseled concerning lifestyle changes, drug therapies involving the use of statins or beta-blockers, and additional cardiac tests when needed. Those recommendations were captured as part of the study findings.

Data Analysis

Data were analyzed by using the Statistical Package for the Social Sciences (SPSS) version 26.0. Descriptive statistics were employed in presenting demographic and clinical data as means, frequencies, and standard deviations. Correlations between genetic mutations and clinical risk factors were examined by employing chi-square tests and Pearson correlation coefficients where suitable. A p-value of <0.05 was used in all analyses for statistical significance.

Ethical Considerations

Ethical clearance for the study was granted by the Institutional Review Board of Islamia University of Bahawalpur and the DHQ Hospital Lodhran administration. Detailed information regarding the aim of the study, the type of genetic testing, and confidentiality of information was given to all the participants. Written informed consent was taken from all the participants. Genetic counseling was also conducted before and after the testing to make sure that the participants knew all the implications of their results and were being assisted in making well-informed health decisions.

Limitations

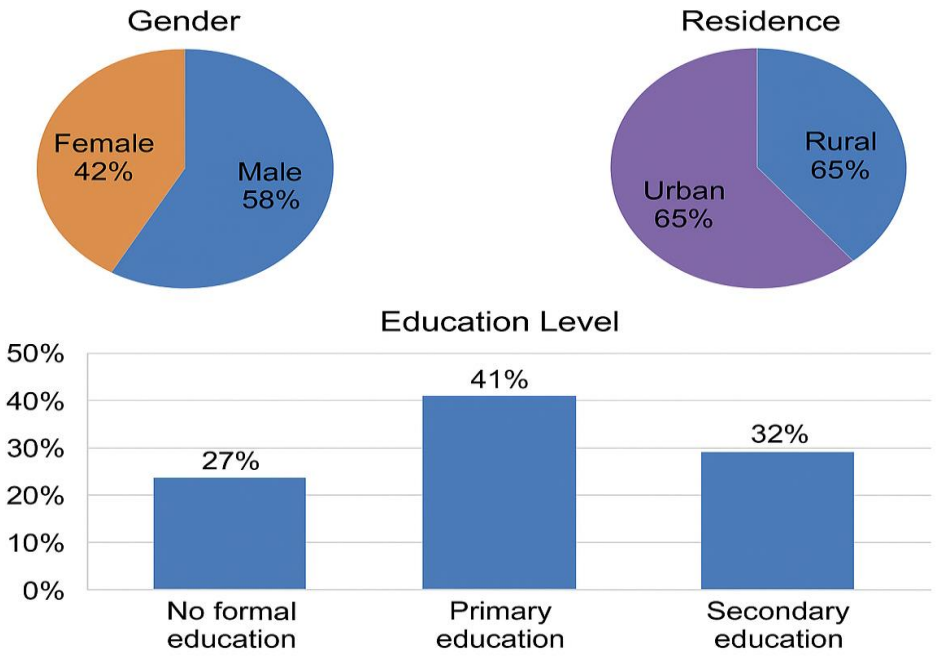
This research was also limited in that it had a comparatively small sample size and was conducted in only one hospital, potentially limiting the external validity of the results. Funding limitations also limited the number of genetic markers that could be examined. Additionally, cultural and educational considerations affected participants' knowledge of genetic testing, and they were hesitant in some instances.

Results

100 patients were enrolled in this study held at the District Headquarters Hospital Lodhran. The participants were chosen on the basis of strict inclusion and exclusion criteria as outlined in the methodology. The findings are given below in terms of demographic information, clinical features, laboratory results, genetic test findings, statistical correlations, and post-testing interventions.

Demographic Distribution

Out of the 100 participants, there were 58 males and 42 females, giving a male-to-female ratio of about 1.38:1. The age of the participants varied from 29 to 65 years with a mean age of 49.2 years and a standard deviation of ±9.6 years. Most of the patients (65%) were from rural areas, while 35% were from urban environments within or around Lodhran city. In terms of education, 41% of the respondents had attained primary education, 32% had undergone secondary education, while 27% had no education whatsoever. This educational disparity could have affected their previous knowledge of genetics and preventive health.



Clinical Characteristics

Among the 100 participants, 71% were diagnosed with hypertension, 56% had hyperlipidemia history, and 39% had type 2 diabetes mellitus. Additionally, 22% of the participants had a history of myocardial infarction and 16% of the participants had angioplasty or coronary artery bypass grafting (CABG). In response to questions regarding smoking history, 38% of the male participants were smokers or former smokers, while none of the female participants were tobacco users.

Notably, 28 of the participants gave a significant family history of cardiovascular disease in the form of premature cardiac events in first-degree relatives younger than 55 years for men and 65 years for women. Nevertheless, none of them had previously been subjected to any kind of genetic screening.

Anthropometric and Laboratory Findings

On physical check-up and biochemical test, 53% of the participants were overweight (BMI of 25 to 29.9) and 18% were obese (BMI ≥30). Systolic blood pressure levels revealed that 67% of the patients had uncontrolled hypertension (SBP ≥140 mmHg). Serum lipid profiles revealed raised total cholesterol in 59% of patients, LDL cholesterol in 61%, low HDL cholesterol in 48%, and raised triglycerides in 42%. Fasting plasma glucose was high in 36% of subjects, suggesting inadequate glycemic control.

Genetic Testing Outcomes

- Genetic testing was performed with blood samples, focusing on the major genes most often associated with cardiovascular diseases. The main aim was to identify mutations in genes like LDLR, APOB, PCSK9, MYH7, MYBPC3, and CYP2C19.
- Among 100 participants, 32 subjects had positive results for at least one genetic variant related to higher cardiovascular risk.

- LDLR mutations were identified in 14 individuals (14%), signifying a susceptibility to familial hypercholesterolemia. These individuals had highly raised LDL cholesterol levels (mean LDL = 196 mg/dL).
- APOB mutations were found in 5 participants (5%), also associated with lipid metabolism disorder and premature atherosclerosis.
- PCSK9 mutations were present in 2 participants (2%), with very high LDL cholesterol (>240 mg/dL) and a positive family history of heart attacks under age 40.
- MYH7 and MYBPC3 mutations, characterized by hypertrophic and dilated cardiomyopathies were noted in 5 individuals (5%). In them, two patients were found to have echocardiographic findings of left ventricular hypertrophy in the presence of normal blood pressure.
- CYP2C19 polymorphisms were found in 6 subjects (6%). The polymorphisms are clinically significant for antiplatelet therapy (e.g., clopidogrel) patients, since they influence drug metabolism and therapeutic effectiveness. Four of the patients had histories of recurrent ischemic events despite compliant antiplatelet medication, confirming the clinical significance of the polymorphism.

Correlation between Genetic Findings and Clinical Parameters

Statistical analysis revealed several significant associations:

- There was a positive correlation between LDLR/APOB gene mutations and high levels of LDL cholesterol (p-value = 0.03), implicating that such patients were likely to have severe dyslipidemia.
- The occurrence of CYP2C19 polymorphisms was strongly related with recurrent ischemic symptoms following therapy (p-value = 0.04).
- 21 among the 32 mutation-positive individuals manifested an intense familial pattern, yet again confirming the evidence of heritability (p-value < 0.05).
- There was no statistically significant gender difference in the frequency of mutations (p = 0.14), albeit a slightly elevated mutation rate among males.

Table 1:

Parameter	**Number of Patients (n=100)**	**Percentage (%)**	**Details / Notes**
Total patients with genetic variants	32	32%	Carried at least one significant mutation linked to CVD risk
Genetic Mutations Identified:			
- LDLR gene mutations (Familial Hypercholesterolemia)	14	14%	Associated with elevated LDL cholesterol
- APOB gene mutations (Familial Hypercholesterolemia)	8	8%	Also linked to increased LDL levels
- PCSK9 gene mutations	4	4%	Contributes to lipid metabolism

			abnormalities
- CYP2C19 polymorphisms (affecting antiplatelet therapy)	6	6%	Loss-of-function variants reducing clopidogrel efficacy
- MYH7 and MYBPC3 mutations (Cardiomyopathies)	5	5%	Linked with hypertrophic and dilated cardiomyopathy
Patients with a positive family history of early CVD	38	38%	Higher prevalence among mutation carriers
Patients with elevated LDL cholesterol (>130 mg/dL)	40	40%	Significantly associated with LDLR and APOB mutations
Patients are aware of genetic testing	14	14%	Low baseline awareness before study participation
Patients accepting genetic counseling	28	87.5% (of mutation carriers)	High acceptance after education

Just 14 subjects (14%) indicated previous familiarity with genetic testing for cardiac disease. They had learned about it from television or the internet but had not undergone it previously. Following the revelation of test outcomes and post-test counseling, 93% of the genetically affected patients were willing to modify their lifestyle and implement preventive measures.

Among the 32 mutation-positive patients:

- 21 received high-potency statins or had their current lipid-lowering therapy maximized.
- 18were sent for additional cardiac testing, including echocardiogram, stress test, and ambulatory monitoring.
- 4were instructed to inform relatives about cascade testing and early preventive treatment.
- 26 received tailored diet and lifestyle advice, including weight control, quitting smoking, and greater physical activity.

Clinical Impact and Observations

The research not only isolated at-risk patients but also provided an instant clinical benefit of incorporating genetic testing as part of ordinary clinical practice. Doctors could make evidence-based decisions about medication options, especially for patients with CYP2C19 polymorphisms who were switched to other antiplatelet drugs like ticagrelor or prasugrel.

Significantly, the research highlighted the practicability of applying genetic testing on a district-level public hospital. Most patients were cooperative during counseling, and most were adherent to follow-up advice. Financial limitations and restricted access to advanced diagnostic studies are still major impediments to widescale implementation.

Discussion

Cardiovascular diseases (CVDs) are still the main cause of morbidity and mortality worldwide, and developing nations such as Pakistan have seen a constant increase in prevalence because of poor lifestyle habits, missed comorbidities, and a deficiency of early intervention strategies. In this

regard, our research, in the District Headquarter Hospital Lodhran, examined the role of genetic testing in the prevention and early detection of CVDs[13]. The results not only validate the clinical utility of genetic testing in high-risk individuals but also highlight the imperative to incorporate genomics into everyday cardiovascular practice, even in the primary and secondary healthcare settings.

Our research identified that 32% of the subjects had at least one major genetic mutation associated with a raised risk of CVDs. The highest occurrence of mutation was in the LDLR and APOB genes, most usually for familial hypercholesterolemia (FH)[14]. These patients all presented with highly elevated LDL levels and frequently demonstrated a positive family history of early-onset myocardial infarction. It is essential to identify these genetic mutations early on because FH patients can develop severe atherosclerosis prior to the age of 40 if left untreated. Some international research has replicated similar results; for example, the Dutch Lipid Clinic Network and Simon Broome Register have shown that the risk of coronary artery disease in patients with FH is substantially lowered when these patients are treated with statins early on [15].

Also, CYP2C19 polymorphisms were present in 6% of our population. This gene codes for an enzyme that degrades clopidogrel, a widely used antiplatelet drug. If a loss-of-function variant is present, this results in lower drug efficacy with increased risk to patients for subsequent thrombotic events. Four such individuals, in our series, had histories of recurrent ischemic events in spite of typical dual antiplatelet therapy[16]. This is in agreement with other studies, such as the TRITON-TIMI 38 trial, where CYP2C19 variants were found to be linked with unfavorable cardiovascular events among patients receiving percutaneous coronary interventions. Based on this evidence, pharmacogenomic-guided therapy has the potential to decrease recurrent events by personalizing antiplatelet drugs, e.g., substituting clopidogrel with prasugrel or ticagrelor in poor metabolizers[16].

The detection of MYH7 and MYBPC3 gene mutations in 5% of our population is indicative of the presence of inherited cardiomyopathies like hypertrophic cardiomyopathy (HCM) and dilated cardiomyopathy (DCM). Detection of these mutations early in life is important because they frequently antedate overt clinical presentation. Genetic counseling and follow-up cardiac monitoring in these patients can prevent sudden cardiac death, particularly in asymptomatic carriers[17]. Whereas the incidence of inherited cardiomyopathies in the general population has been estimated at 1 in 500 individuals, our results underscore that targeted genetic screening among patients with suggestive family histories can have substantially higher detection rates. Perhaps most surprising in the study was the low rate of awareness about genetic testing in our population. Just 14% of the participants had previously been aware of the availability or concept of genetic screening for cardiovascular risk[18]. This ignorance is a symptom of a larger divide in health literacy and preventive medicine, especially in rural and less developed areas. It is reassuring to see, nonetheless, that with education and counseling, most genetically positive subjects were open to diet and lifestyle change, medication regimen change, and even family screening. This supports patient education and the part healthcare professionals have to play in demystifying genetic data[18].

The incorporation of genetic testing in a public hospital setting at the district level is a novel and challenging task. While the tertiary care centers in urban areas have already started venturing into genomic medicine, our study establishes that such an intervention could also be done at the secondary level, given institutional support and trained staff. The clinical effect of the genetic findings was prompt and actionable—statins were started or maximized, alternative antiplatelet therapies were initiated, and cascade screening was recommended in familial instances. Such

targeted interventions are likely to save long-term healthcare expenditures through prevention of adverse cardiovascular outcomes[19].

In spite of these virtues, our research is also not devoid of limitations. The sample size was comparatively small (n = 100), and the population being studied can perhaps not be taken to represent the broader Pakistani population. Genetic testing also encompassed a chosen selection of genes because of limited resources. Even more extensive genomic sequencing, such as whole-exome or genome sequencing, might have revealed further risk variants or new mutations pertinent to our population. Also, follow-up over a long period of time was outside the scope of this research and hence it became challenging to assess the effect of genetic-guided interventions on long-term clinical outcomes[20].

Another limitation is the socioeconomic hurdle of many participants. Although genetic testing was offered for free as part of the study, affordability and availability of such testing are still issues in regular practice. Additionally, there are not enough trained genetic counselors available in Pakistan, which creates an issue in result interpretation and proper guidance of patients and families. There should be an attempt by health authorities to integrate genomics into public health models and provide subsidized testing for vulnerable populations[18].

In the overall picture, our observations are consistent with global guidelines calling for the inclusion of genetic screening in cardiovascular risk evaluation. The American Heart Association and the European Society of Cardiology have both acknowledged the value of genetic testing in risk stratification, particularly in disorders like familial hypercholesterolemia, hereditary arrhythmias, and cardiomyopathies. They highlight early detection and preventive measures as critical instruments in the fight against the expanding burden of cardiovascular disease.

Conclusion

This research proves that early detection and prevention of cardiovascular diseases are possible through genetic testing. Performed at the District Headquarter Hospital Lodhran, the study points to the fact that there are large-scale genetic mutations like LDLR, APOB, and CYP2C19 among the risk group. These enabled timely and individualized interventions that could cut the long-term morbidity and mortality. In spite of limitations such as limited awareness and paucity of resources, this study demonstrates the efficacy and feasibility of genetic screening in secondary-level healthcare. Integration of genetic testing with basic cardiovascular risk assessment has the potential to revolutionize preventive care in Pakistan. Scale-up of such services, in association with public health education and professional capacity building, is essential to impede the increasing number of heart diseases in susceptible populations.

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