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Compulsory pre-marital thalassemia screening to mitigate the burden of thalassemia major on society and healthcare system of Pakistan

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ABSTRACT

Thalassemia imposes a significant burden on both society and the healthcare system of Pakistan, with high prevalence rates and substantial financial implications. Pre-marital thalassemia screening emerges as a crucial strategy to alleviate this burden by identifying carriers early and enabling informed reproductive choices. Despite the evident need, Pakistan currently lacks infrastructure and awareness for such screening programs. This paper advocates for compulsory pre-marital thalassemia screening as a proactive measure to reduce the incidence of thalassemia major and its associated healthcare costs. Drawing on successful models from other countries, it proposes an action plan involving policy implementation, public awareness campaigns, healthcare infrastructure development, and professional training. By addressing challenges such as infrastructure gaps, societal misconceptions, and ethical considerations, Pakistan can effectively combat the spread of thalassemia, thereby enhancing public health outcomes and alleviating the strain on healthcare resources.

Keywords. Thalassemia, pre-marital screening, Pakistan, Healthcare burden.

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INTRODUCTION

The implementation of pre-marital thalassemia screening is significant to mitigate its negative impact on society and the healthcare system. Worldwide, 1.5% of individuals are thalassemia carriers, 399 million thalassemia carriers were estimated in 2019.[1,2] There are more than 10 million carriers of the beta thalassemia trait in Pakistan, whereas the prevalence of the condition ranges from 5 to 8%.[3] Each year, about 5000 children are diagnosed with thalassemia major in Pakistan.[4] The average life expectancy of a thalassemia major patient is 10 years in Pakistan.[5] The population of Pakistan has an estimated 9.8 million beta-thalassemia carriers. The beta thalassemia gene is present in about 6% of Pakistani people.[6] Pakistan has 50,000 registered thalassemia patients which causes a significant burden on the health system.

Thalassemia major patients require frequent blood transfusions and Pakistan has one of the highest rates of transfusion-dependent thalassemia patients globally.[7] Approximately 2000 children are reported with beta-thalassemia in Baluchistan.[8] According to estimates, 5000–9000 children in Pakistan are born each year with beta-thalassemia major, with a carrier rate of 5–13%. In the Punjab province, there was only one government-funded project, the Punjab Thalassemia Prevention Program which offered free services for beta thalassemia screening and prenatal diagnosis.[9] Moreover, patients with thalassemia major are at great risk of developing complications such as liver cirrhosis and cardiac issues because blood transfusion is the only available treatment, and most patients lose their lives from iron overload at the age of 30 or sooner.[8] Therefore, this position paper supports the compulsory pre-marital thalassemia screening to mitigate its detrimental impact on society and the healthcare system of Pakistan.

Position Standpoint

Thalassemia exerts immense strain on the society and the healthcare system of Pakistan. The financial association related to the diagnosis, treatment, management, and long-term care of patients imposes a significant burden on families and the healthcare system. Pre-marital thalassemia screening is helpful to mitigate the burden of the disorder and to promote the health and well-being of the public. Therefore, pre-marital thalassemia screening needs to be compulsory to effectively reduce its negative impact on the society and healthcare system of Pakistan.

Background

There are multiple reasons for the high prevalence of thalassemia in Pakistan. First of all, genetic mutation; second, high birth rate; third, consanguineous marriage custom.[7] Social and cultural factors such as low-income status, lack of awareness, and marriages in the same ethnic groups are also contributing to the high frequency of the disease.[10] The high prevalence of consanguineous marriages in Pakistan is the most significant cause of thalassemia. Pakistan has a significantly high rate of consanguineous marriages. Over the past three decades, there is a 63% increase in this rate.[11] Pre-marital screening is a valuable tool to identify

this genetic disorder. Many Muslim countries (Saudi Arabia, Palestine, and Cyprus) have adopted mandatory pre-marital scree approaches to prevent the burden of the disease and to secure the future of the next generation.[12] Currently, Pakistan has no infrastructure for premarital screening and disease prevention.

Thalassemia is an inherited autosomal recessive disorder in which blood does not have enough hemoglobin. It leads to malfunctioning and defective red blood cells and causes anemia.[10] The patient with thalassemia can have mild or severe anemia that in the long-term causes damage to multiple organs and often leads to death. After sickle cell disease, it is the second-most prevalent hemoglobinopathy globally.[10] It is a hematological disorder that can be classified based on two factors including specific parts of affected hemoglobin (alpha and beta) and the severity of the disorder (trait, carrier, intermediate, major). There are two types, major and minor thalassemia. Minor thalassemia includes mutation of one gene and demonstrated mild symptoms, while major thalassemia includes mutation of two or more genes and demonstrated moderate to severe symptoms. Patients with thalassemia major need frequent blood transfusions; thus, iron chelation therapy is essential for treating post-transfusion iron excess.[13]

The Burden of Thalassemia and Implications for Healthcare

Thalassemia poses a remarkable economic burden not only on affected families but also on the healthcare system. It negatively affects the quality of life of patients, and their families. It reduces the productivity of affected individuals and demands high costs for continuous medical management. Since access to the only treatment, that is bone marrow transplant, is not accessible for most people due to its huge financial cost and compatibility issue of bone marrow, the only option left in most cases is ongoing transfusions and management of complications. It imposes an extra burden on the families and healthcare system of Pakistan.

The average monthly cost for a blood transfusion of a thalassemia patient is Rs. 9626.[1] This expense causes a heavy financial burden on the families. The cost management increases with specialized medical care, iron chelation therapy, and management of complications (splenomegaly, cardiomyopathy, and hepatitis B and C). The cost of treatment for families varies as per their entitlement status and the institution where they are seeking health services. The Fauji Foundation Hospital (FFH) incurs a monthly cost of Rs. 5000 to 10,000, and the Pakistan Institute of Medical Sciences (PIMS) approximately Rs. 80,000 for patients of thalassemia.[14]

As per the economic survey of Pakistan (2022), health expenditure increased by 30% in the fiscal year 2021-22 as compared to 2020-21 from Rs 505.4 billion to Rs 657.2 billion. Public sector expenditure on the healthcare system was estimated at 1.2% of GDP in 2020-2021 as compared to 1.1 percent in 2019-2020 against the 5% recommendation of the World Health Organization.

Pakistan has been the victim of an economic crisis for years and already invest less than required in its healthcare system. Therefore, there is a dire need to address the issue of thalassemia to reduce the burden on the healthcare system of Pakistan.

Significance of Compulsory Pre-marital Thalassemia Screening

Compulsory genetic screening before marriage has the potential to prevent this genetic issue. The pre-marital genetic screening could help to identify individuals with traits of thalassemia. Additionally, early detection facilitates informed decisions about marriage and reproduction. One study demonstrated that only 1.7% of couples with a thalassemic child had pre-marital screening while 98.2% had no screening for thalassemia before marriage.[5] The detection of the thalassemia gene before marriage enables the couple to plan their family intelligently and take the required safety measures to avoid the birth of children with thalassemia major. Prevention of the disease through premarital screening is the best approach to deter marriages between carriers.[15]

Thalassemia major is a chronic condition requiring costly treatment of lifelong blood transfusion and chelation therapy which add a burden on the healthcare system as well as on the families. Early identification of the carriers with premarital testing would result in a reduced number of children born with thalassemia major in turn reducing the burden upon healthcare including the use of resources hospital admissions, specialized care, and continuous therapies. A reduction of up to 70% of thalassemia cases has been achieved in Saudi Arabia in six years, while no affected birth in Cyprus occurred within 5 years of the introduction of the premarital screening program.[16]

In patients with major thalassemia the quality of life is significantly decreased in addition to compromised activities, relationships, and abilities.[17] Patients with thalassemia major require regular blood transfusions which results in multiple medical complications such as organ failure and abnormal growth. Prevention of birth of thalassemic child with premarital screening will help society to improve their quality of life; it will eliminate their emotional, physical, and financial burden, hence improve their quality of life. Pre-marital screening for thalassemia will provide an opportunity for the healthcare professional to support and counsel the individuals carrying the disease. People with carrier status could be provided with genetic counseling and support to learn about the disease and its effects. Genetic counselors would be able to provide them with sufficient knowledge and support in making well-informed decisions about their families.

Gap Identification

There is significant progress in public health genomics policies in developed countries and some Muslim countries, but Pakistan has no sufficient initiative in genetic services.[18] Pakistan has a high prevalence of family marriages but still lacks programs for pre-marital screening, genetic awareness, and counseling. The healthcare system focuses on infectious diseases and de-prioritizes genetic disorders with limited funding for genetic health services.[18] Screening for thalassemia includes complete blood count (CBC), single tube osmotic fragility test, Hb electrophoresis, and DNA analysis. In the case of beta-thalassemia if both parents are thalassemia minor, there is a 25% chance of an affected child during every pregnancy. But still, there is no infrastructure available for pre-marital screening.

Current Policy Initiative Related to Thalassemia Screening in Pakistan

Khyber Pakhtunkhwa, Punjab, and Sindh assemblies have already passed the bill for pre-marital screening of thalassemia, but the majority of the population of Pakistan has insufficient knowledge and alarming low practice of screening.[6] Although other countries have higher awareness levels about the disease, for instance, United Arab Emirates (63%), Thailand (79.6%), Students in Iran (14.7%); other studies showed a reduction in risk marriage rates after proper counseling in Saudi Arabia (10%) and Jordan (40%).[12]

Potential Challenges and Solutions

The unavailability of infrastructure for screening is a challenge to implement a pre-marital thalassemia screening program. There are no thalassemia screening facilities available, especially in rural and remote areas. There is even no infrastructure at the national level to prevent this genetic disorder.[4] But it can be overcome with strong healthcare leadership, political commitment, and social awareness. Iran has demonstrated an example of utilizing existing primary care facilities for thalassemia screening. Pakistan can also adopt this strategy to implement a pre-marital thalassemia screening program.[19]

Lack of awareness and societal acceptance of genetic disorders are also significant barriers to ensuring pre-marital thalassemia screening. Due to a lack of awareness regarding the health risk of cousin marriages, 60% of parents prefer to marry their child to near blood relatives.[11] Public awareness is an instrument for the success of screening programs. The Latium region of Central Italy has successfully decreased the incidence of thalassemia through awareness and screening programs by targeting the school-age population. The attitude and practices of the public regarding pre-marital screening can be changed through education and awareness about thalassemia. The success of the thalassemia screening program depends on effective counseling and genetic education.

Compulsory pre-marital screening also has certain ethical challenges. While keeping the autonomy of a person in view, careful consideration of local beliefs, values, culture, and religious variations is crucial for the implementation of the compulsory screening program. Although international guidelines recommend voluntary screening, some countries implemented compulsory screening and effectively reduced the burden of genetic disorders.[19] Other factors that are an obstacle in the way of pre-marital screening include the culture of consanguineous marriages, fear of a positive result, fear of termination of weddings, societal pressure, fear of inability to find another partner, and fear of social disgrace.[12]

Action Plan for Compulsory Pre-marital Screening in Pakistan

There is a dire need for policy and legislative measures of compulsory pre-marital screening for all couples before marriage registration. Collaboration with relevant healthcare authorities, policymakers, religious leaders, and stakeholders is necessary to make it successful. Secondly, the launch of widespread awareness campaigns to educate the public about this genetic disorder through various communication channels including television, radio, social media, newspaper, and seminars in educational institutions is essential. The role of religious leaders and community leaders to educate the public is significant here to address the cultural and religious misconceptions regarding screening.

Strengthening the healthcare infrastructure is the backbone to make it possible. There is a need to establish pre-marital screening centers across the country, especially in rural and underserved areas, who can collaborate with existing healthcare institutions to conduct screening tests to ensure easy access to screening centers. Also, specialized training sessions for the healthcare professionals for screening and genetic counseling to guide and assist couples in making informed decisions about their marriages and family planning.

CONCLUSION

Thalassemia is a genetic disorder in which blood does not make enough hemoglobin. It exerts immense strain on the society and the healthcare system of Pakistan. Pre-marital thalassemia screening is helpful to mitigate the burden of the disorder and to promote the health and well-being of the communities. Pakistan has a high prevalence of family marriages but still lacks pre-marital screening, genetic awareness, and counseling programs. There is significant progress in public health genomics policies in developed countries and some Muslim countries, but Pakistan has no sufficient initiative in genetic services. The unavailability of a screening infrastructure is a challenge to implement a pre-marital thalassemia screening program. But it can be overcome with strong healthcare leadership, political commitment, and social awareness. It is high time to implement nationwide compulsory pre-marital screening. Through this action, Pakistan can effectively mitigate the spread of thalassemia, reduce its economic burden, and improve the health and well-being of future generations in Pakistan.

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