DOI: https://doi.org/10.54393/pbmj.v7i11.1184

PAKISTAN BIOMEDICAL JOURNAL
https://www.pakistanbmj.com/journal/index.php/pbmj/index
ISSN (P): 2709-2798, (E): 2709-278X
Volume 7, Issue 11 (November 2024)

Thalassemia in Pakistan: Addressing a Genetic Health Crisis

OPEN ACCESS

Tahir jamil[1]

[1]King Abdul-Aziz University, Jeddah, Saudi Arabia

tjahmed@kau.edu.sa

ARTICLE INFO

How to Cite:

Jamil, T. (2024). Thalassemia in Pakistan: Addressing a Genetic Health Crisis . Pakistan BioMedical Journal, 7(11). https://doi.org/10.54393/pbmj.v7i11.1184

Thalassemia, a genetic disorder usually caused by the defective genes. These genes are responsible for production of hemoglobin, a protein necessary for the transfer of oxygen in body through blood. Prevalence of thalassemia in Pakistan is concerning to a significant level. Global prevalence of this disease has affected millions of people and their lifestyles. Moreover, the disease leads to the burden on the diseased person's family and the medical resources of the country, causing an alarming concern.

The main issue of high prevalence of this disorder in Pakistan is associated with high percentage of cousin marriages. This cultural practice between the close relatives increases the chances of inheriting mutated genes into next generations. Each year more than 5000 children in Pakistan are born with disease. The rate of thalassemia is considerably higher in first cousin marriages (76.7%) than in second cousin marriages (23.3%) [1]. Furthermore, the inadequate spread of awareness among people and the lack of preventative measure by people is contributing to the increase in the number of thalassemia cases

Thalassemic patients do require blood transfusion in an adequate to survive and live a normal life. Moreover, after transfusion they need financially draining therapies like iron chelation that is being required to manage the overload of iron in blood. The limited medical resources and poor infrastructure of hospitals and clinics in the rural areas can lead to the increase of complications then reducing the problems come with this disease. This kind of public healthcare system can affect the diseased individuals to level to develop life threatening problems such as stunted growth, and organ failures etc.

To address this problem, a national program stating that genetic screening before marriage is necessary can lead to the reduction in the rate if this disease. Premarital screening for inherited diseases including Thalassemia is being carried out in middle east for last more than a decade. Some countries like Iran have overcome this problem to a great level by taking these kinds of initiatives, which shows that how important is it to detect the disease occurrence through premarital screening and counseling [2]. There should be campaigns for the public awareness highlighting the importance of genetic screening so that the cause of occurrence of this disorder can be decreased.

Additionally, medical advancements have provided the hope. Despite being costly and scarce, for certain patients with thalassemia, bone marrow transplants (BMT) have proven to be a curative treatment, success rates for those who satisfy the right criteria, such as younger patients with matched related donors, range approximately between 80 and 95% [3]. Future advances in repairing genetic abnormalities at their origins may be possible thanks to emerging gene-editing technologies like CRISPR. To make these remedies available in Pakistan, however, significant funding for medical infrastructure and research is needed.

Despite being avoidable, thalassemia will continue to take lives and put a burden on families and communities unless immediate action is taken. Pakistan can considerably lessen the impact of this genetic health disaster and clear the path for a healthier future by placing a high priority on education, early identification, and cutting-edge treatments.

REFERENCES

[1] Masih N, Amir F, Tabbasum R, Naz A, Nadeem A. An Empirical Investigation of the Relationship Between Consanguineous Marriage and Prevalence of B-thalassemia in Punjab, Pakistan: A Cross-sectional Study. International

DOI: https://doi.org/10.54393/pbmj.v7i11.1184

journal of health sciences.;7(S1):2362-7.
[2] Khorasani G, Kosaryan M, Vahidshahi K, Shakeri S, Nasehi MM. Results of the national program for prevention of β-thalassemia major in the Iranian Province of Mazandaran. Hemoglobin. 2008 Jan 1;32(3):263-71.
[3] Hussein MH, El Missiry M, Khalid S, Yaqub N, Gilani SK, Fatima I, Zara T, Marwah P, Soni R, Bernard F, Manna A. Bone marrow transplantation for thalassemia: a global perspective. Thalassemia Reports. 2013 Mar;3(s1):e42.