

Conference Abstract



Thalassaemia Prevention: Let's Do the Doable

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Cite this article as: Bashir S. Thalassaemia Prevention: Let's Do the Doable. Ann Pak Inst Med Sci. 2024;20(Suppl. 2):922. doi: 10.48036/apims.v20iSuppl.2.1297.

Thalassaemia is a major public health challenge in Punjab, impacting thousands of families each year. Characterized by a genetic defect in haemoglobin production, thalassaemia leads to severe anaemia and requires lifelong management, including regular blood transfusions and iron chelation therapy. The prevalence of thalassaemia in Punjab, fueled by high rates of consanguineous marriages, remains a significant concern.

Punjab has made strides in addressing thalassaemia through a range of preventive and supportive measures. Punjab Thalassaemia and Other Genetic Disorders Prevention & Research Institute (PTGD) is an initiative by the Specialized Healthcare & Medical Education Department, Government of Punjab, aimed at reducing Thalassaemia Major births in Pakistan. Established in line with the Millennium Development Goals (MDGs) and the government's vision, PTGD addresses the high prevalence of genetic disorders, notably beta thalassaemia, which affects 6% of the population. With no effective treatment available, prevention program, modeled after successful strategies in Cyprus and Iran, are essential. The Punjab Thalassaemia Prevention Programme, launched in 2009-2010, achieved notable success and was expanded in July 2021 to include other genetic conditions, transitioning into a permanent institute under Fatima Jinnah Medical University. The institute now offers comprehensive prevention and screening services across Punjab. Despite the preventable nature of this disorder, a coherent national policy is lacking, leading to severe impacts on patients, families, and an already strained healthcare system. The current efforts, primarily led by NGOs and partially supported by the government, are insufficient.

Prevention is the only viable solution, proven cost-effective through global models in countries like Iran, Turkey, and Greece. It advocates strategies such as awareness campaigns, extended family screening, premarital and population screening, and prenatal diagnosis via chorionic villus sampling (CVS). Despite these efforts, challenges persist. Awareness in rural and underserved areas is limited, leading to inadequate participation in screening programs. Additionally, many families face difficulties in accessing comprehensive care, particularly in remote regions where specialized medical services are scarce. Funding constraints and insufficient infrastructure also impede consistent and effective implementation of preventive strategies. There is an urgent need for collaborative efforts and sincere discussions to establish effective, cost-efficient strategies. A committee is needed to conduct meta-analyses and audits, recommending joint actions to be presented at both federal and provincial legislative levels.

Government and non-governmental organizations have collaborated to launch awareness campaigns to educate the public about the disease and its genetic implications. Mandatory premarital screening programs are being promoted to identify carriers of the thalassaemia gene. Several hospitals and dedicated centers are now equipped with specialized facilities to diagnose and manage thalassaemia patients. Moving forward, comprehensive strategies are required to strengthen thalassaemia prevention in Punjab. Expanding public health campaigns, making premarital screening mandatory, and enhancing access to genetic counseling are pivotal steps.

Disclosure

The author(s) declare no conflicts of interest. This study was presented as an oral presentation during the 5th International Annual Conference of BBMT-Pakistan (Bring Brilliant Minds of Transfusion) in Langkawi, Malaysia, December 5-6, 2024. The abstract is published in Annals of PIMS. 2024;20 (Suppl. 2; doi: 10.48036/apims.v20iSuppl.2.1297).