

Prenatal Diagnosis and Prevention of Thalassaemia in Punjab, Pakistan

Prof. Noreen Akmal, MBBS, FCPS, Masters in Maternal Fetal Medicine

Visiting Professor, Fatima Jinnah Medical University Lahore, Pakistan.

Dr. Yasmeen Ehsan, Director General Punjab Thalassaemia and other Genetic Disorders Prevention and Research Institute

Introduction

Pakistan is the 5th most populous country in the world with a population of approximately 250 million. In Pakistan β -thalassaemia (β -thal) trait frequency ranges between 5.0–7%. There are more than 10 million carriers in the country; and every year, around 5000-8000 children are born with (β -TM) β -thal major (1). No standard management protocols exist and blood transfusion remains the mainstay of management.

Currently in Pakistan, at the national level, a comprehensive Thalassaemia Prevention Program does not exist. However, at the provincial level, the Government of Punjab took the lead and initiated Punjab Thalassaemia Prevention Programme (PTPP) in 2009-2010. **The PTPP has been very successful and is one of the world's largest prevention programme.** Based on its success, the Punjab Health Department decided to enhance the scope of the project to other genetic conditions as well. In July 2021 the PTPP was converted into an ongoing program titled “Punjab Thalassaemia and other Genetic Disorders Prevention and Research Institute (PTGD)” as an affiliated unit under Fatima Jinnah Medical University, Lahore.



Punjab Thalassaemia Prevention Program

The institute's head office is in Lahore with nine regional centers situated in all the nine divisional headquarters of Punjab Province. The institute has a community outreach component. In each district there is a mobile field team, which provide targeted screening (extended families of affected individuals) as well as premarital screening services to general population at their doorsteps.

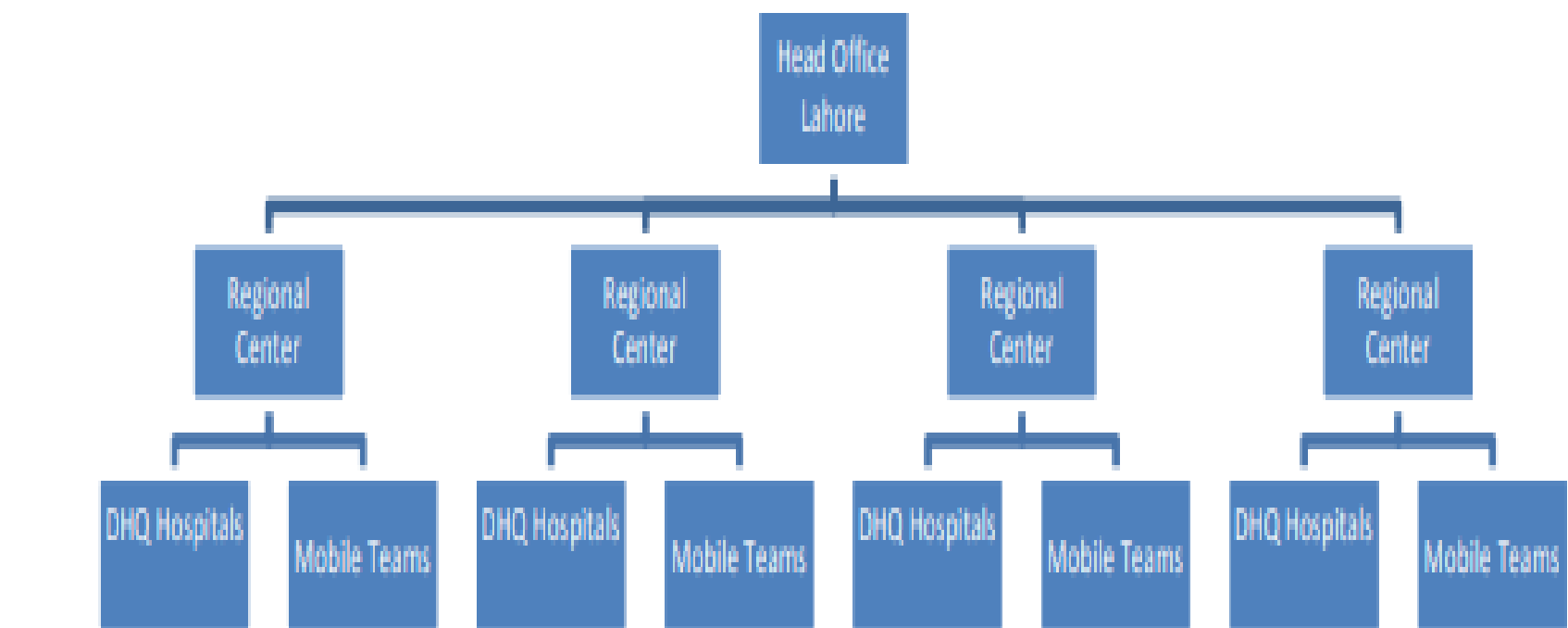
Approximately more than 40,000 individuals have been screened as part of extended family screening. Genetic counselling is provided to these individuals according to the test results so that they can make informed decisions about marriages and prenatal diagnosis.

Haematology services

Each regional center has a haematology Laboratory providing free facility of beta Thalassaemia carrier screening. These laboratories are equipped with latest and fully automated CBC Analysers (Sysmex), Capillary II Zone Gel Electrophoresis, and High-Performance Liquid Chromatography (HPLC).

Prenatal Diagnosis

- Prenatal diagnosis is provided to couples who are carriers of beta thalassaemia minor. Trans-abdominal Chorionic Villus Sampling (CVS) is done between 11-15 weeks of gestation.
- **So far 10,579 CVS procedures have been performed since June 2012 to December 2024. Out of these 2,696 (25.48%) were Thalassaemia Major, 5,253 (49.65%) were carriers and 2,630 (24.86%) were normal.**
- Procedure related miscarriage rate is 1.001%.
- CVS test facility is provided at all Regional Centers free of cost.
- The couples are counseled before and after the test to make sure they understand why they are taking the test and to receive professional guidance about the choices they have in managing the pregnancy.



PUNJAB THALASSAEMIA & OTHER GENETIC DISORDERS PREVENTION & RESEARCH INSTITUTE

Government of the Punjab



DNA Laboratory

The PTGD has established a DNA Laboratory at Sir Ganga Ram Hospital, Lahore, which is the first public sector DNA Lab providing free genetic testing and prenatal diagnosis services for patients of Thalassaemia. The samples are analyzed by PCR based Primer- specific Amplification method.



Genetic counselling

PTGD has trained genetic counsellors in all regional centers. Non directive counselling is provided in accordance with moral, ethical and religious values. If the fetus is diagnosed to have Thalassaemia Major, the parents are informed regarding the lifelong implication of this debilitating condition and the treatment options available. They are also informed about Islamic stance on the termination of affected pregnancies before 120 days of gestation, as it is the time when the soul is given to the unborn child. The couple can then decide regarding termination or continuation of pregnancy. The best possible solution is offered to couples while strictly maintaining their confidentiality



Conclusion

- Beta thalassaemia remains a significant public health challenge in Pakistan. Managing the condition imposes a substantial economic burden on families and the healthcare system. Therefore, the most effective strategy is to focus on prevention through comprehensive screening and early diagnosis.
- Trans-abdominal chorionic villus sampling (TA- CVS) is a safe and feasible prenatal diagnostic procedure.
- Providing accurate prenatal diagnosis alongside comprehensive genetic counselling, which also addresses the religious perspective, can change views on prenatal diagnosis and selective pregnancy termination.
- **National Level Thalassaemia prevention program, by providing diagnostic services in adults and prenatal diagnosis in carrier couples can lead to reduction in the disease burden.**

Reference

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Acknowledgements and Contact details

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Contact details:- Prof. Noreen Akmal

Cell No:- +923009430531

Email:- noreenakmal@fjmu.edu.pk