

# Thalassemia: What Do We Know?

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## Key points

- Thalassemia Prevalence in Pakistan.
- Transfusion transmitted infections.
- How to Raise awareness.

Thalassemia, a genetic condition, is characterized by impaired production of globin chains, which leads to anemia and ineffective erythropoiesis. There are three main types of thalassemia: thalassemia major, thalassemia intermedia, and thalassemia minor. Thalassemia major is the most severe form and requires frequent blood transfusions. Beta-thalassemia specifically involves reduced synthesis of the beta chains of hemoglobin, resulting in varying phenotypes ranging from severe anemia to normal individuals. Beta-thalassemia is highly prevalent in the Middle East, India, Pakistan, and Southeast Asia.<sup>1</sup>

Thalassemia is the most common hereditary disorder in Pakistan. The trait frequency ranges from to 5-7 % (8-10 Million), which means that there are 160 million carriers. Unfortunately, this number is increasing by 5000 to 9000 every year. One of the major causes of thalassemia in Pakistan is the Cousin marriages. Consanguinity marriage is prevalent among most  $\beta$ -thalassemia parents. Additionally, the high incidence of thalassemia in rural areas can be attributed to low literacy levels and limited awareness of the disease among the population. This lack of knowledge increases the risk of thalassemia in individuals.<sup>4</sup>

Proper counseling of families affected by thalassemia can help dispel misconceptions and superstitions often associated with the disease, particularly among the illiterate population. It is essential to implement screening programs in schools, colleges, and

universities to detect disease carriers. Moreover, raising awareness of thalassemia through seminars and social media can play a significant role in educating people about the disease and its prevention.<sup>4</sup>

Thalassemia patients often need blood transfusion, which significantly increases their likelihood of acquiring transfusion-transmitted infection (TTI).<sup>3</sup> Therefore, the mortality rate of children with thalassemia major is high, as blood transfusion services are quite fragmented due to a lack of resources, and the government is unable to afford better facilities for these patients. To date, 1800 blood centers are functional in the country that are mostly run by private organizations. These private organizations are unable to maintain hygienic conditions due to a lack of funds, which leads to further complications for the patient in the form of transfusion-transmitted infections (TTIs) such as Hepatitis C, Hepatitis B,

HIV, and Syphilis.<sup>2</sup>

Furthermore, due to the unhygienic conditions frequent transfusion of blood to thalassemia patients significantly increases their likelihood of acquiring transfusion-transmitted infection (TTI). Hence, targeting low-risk donors through a screening questionnaire, ensuring an adequate supply of quality screening tests, and conducting awareness campaigns regarding TTIs are necessary measures to further decrease the risk of transmission of these infections in Pakistan.<sup>3</sup>

In conclusion, Proper counseling of families prone to be affected by thalassemia, can help dispel misconceptions and superstitions often associated with the disease, particularly among the illiterate population. It is essential to implement screening programs in schools, colleges, and universities to detect disease carriers. Moreover, raising awareness of thalassemia through seminars and social media can play a significant role in educating people about the disease and its prevention.<sup>4</sup>

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