

Management of Thalassemia in Pakistan

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Beta Thalassemia is the most common genetic disorder. The disorder is prevalent worldwide. About 3% of the world's population carries the genes for Beta Thalassemia and it is estimated that every year about 60000 thalassemic babies are born all over the world.¹ Approximately 79% of affected births are in the Asian population. Carrier rate in Pakistan ranges between 5-8%, and around 5000 children are diagnosed each year with beta thalassemia in Pakistan.² Consanguinity is the main factor leading to high prevalence in Pakistan. There are 25,000 children registered with thalassemia federation of Pakistan however the actual figure is much higher which may be around one lac, as a lot many are living in villages that are not registered with any thalassemia center.

The main components of management of thalassemia include 1. Conservative management which take into account, medical consultation, lab investigations, safe blood transfusion, chelation therapy, treatment with HbF augmenting agents, 2. Curative management comprising of bone marrow transplant and 3. Preventive management comprising of thalassemia screening and genetic counselling. Accomplishment of ideal management thus requires multidisciplinary team addressing all these aspects. However, very few centers furnish for all these components of optimal thalassemia management under one roof. Management of Thalassemia in developing countries like Pakistan poses a major challenge. There are more than 40 Thalassemia centers across the country. Majority of them are giving only transfusional support. Government does not provide required support and majority of burden is borne by NGOs. The main stay of conservative management is blood transfusion, iron chelation, management of complications of iron overload, and prevention. Frequent blood transfusion leads to iron overload requiring treatment with iron chelating agent Serum ferritin level vary widely and could not be controlled due to poor compliance to chelation. The main reason for this noncompliance is the cost of treatment, that is beyond the reach of the common man. Majority of patients thus have high ferritin level, and present with complications of iron overload. Pakistan Baitulmal is providing some support in this regard but majority of patients remain unaddressed.

As far as transfusion therapy is concerned, majority of centers are trying to fulfil needs of patients but there are

reservations about screening methods. The well-functioning blood bank is of primary importance for the management of thalassemia. Its functions are not only to provide the regular supply of blood for the treatment of thalassemia, but also to ensure supply of safe blood, in order to minimize the risk of transfusion-transmitted infections. Various studies have shown that 30-40% thalassemics show seropositivity for HCV and about 4-6 % become HBV positive, despite the availability of vaccine for Hepatitis B.³⁻⁵ Though, some studies have reported that for the past 10-12 years the better screening methods in blood banks in Pakistan have significantly reduced the frequency of these diseases. Further improvements need to be done to implement uniform screening of blood throughout the country.⁶ Another big dilemma is that, many of the private centers do not have a full time treating physician/clinical hematologist. As regards curative aspect, there are very few centers (public and private) throughout the country that are offering bone marrow transplant and due to high cost, it is out of reach of majority of people.

Regarding preventive aspects, awareness and availability of antenatal diagnosis is limited. The main preventive strategies for thalassemia include appropriate information of the disease and importance of screening through awareness programmes, screening and counselling of target families, premarital and prenatal screening. Premarital and prenatal screening have significantly reduced the prevalence of β Thalassemia in various countries as Italy, Greece, Cyprus, Iran, and Saudi Arabia with significant reduction in births of affected children⁷ In Pakistan premarital screening and even antenatal screening is almost nonexistent even in urban areas. Some hospitals have started with antenatal screening and prenatal diagnosis but still due to economic constraints most of the couples are underprivileged.

There is thus a dire need for dedicated thalassemia unit in all big hospitals that oversees all aspects of treatment and where patients may be referred to the specialists whenever required. The centre should offer free medical consultation, genetic counselling, specialized pathology investigations, safe blood transfusion and chelation therapy to the patients. There should be multidisciplinary team comprising of Hematologist, Pediatrician, Cardiologist, Endocrinologist, Psychiatrist/Psychologist, Social worker and dedicated and trained staff. The unit should also try to establish bone

marrow transplant unit and offer this facility at minimum bearable cost. In nutshell an ideal thalassemia centre should have three main units; one dealing with diagnosis and treatment, having close liaison with pediatrician, endocrinologist, cardiologist, psychologist etc. One unit focusing on preventive measures including screening and genetic counselling programme and the third one bone marrow transplant unit. The centre should also have collaboration with national and international organizations. The centre should aim at providing greatest possible level of comfort and convenience for patients and their families and ensure their long and quality life.

References

1. Eleftheriou A. About thalassemia. *Thalassemia Int Pub* 2003; 2: 11-2.
2. Ansari SH and Shamsi TS. Thalassaemia Prevention Programme. *Hematology updates* 2010;23-28 3. Rehman A. Beta Thalassaemia Prevention and Pakistan *Pak Paed J* 2011; 35(2): 55-62.
3. Khattak I, Shah M , Ahmed I, Rehman A , Sajid M. frequency of hepatitis B and hepatitis C in multitransfused beta thalassaemia major patients in district Swat. *Journal of Saidu Medical College* 2013; 3(2).
4. Hussain H, Iqbal R, Khan MH, Iftikhar B, Aziz S, and Burki FK et al. Prevalence of hepatitis C in beta thalassaemia major.. *Gomal Journal of Medical Sciences* 2008;6(2): 87-90
5. Ali MR, Arif M, Arif A and Fatima T. Viral Hepatitis C in Thalassaemia: Determination of Antibody HCV Frequency in Mutitransfused Thalassaemia Patients *A.P.M.C* 2016;10 (1): 20-25.
6. Saqib H. Ansari, Tahir S. Shamsi, Mohammed Tahir Khan, Kousar Perveen, Tasneem Farzana, Sajida Erum and Iqra Ansari Seropositivity of Hepatitis C, Hepatitis B and HIV in Chronically Transfused β -Thalassaemia Major Patients *Journal of the College of Physicians and Surgeons Pakistan* 2012; 22 (9): 610-611
7. Baig SM, Sabih D, Rahim MK and Azhar A et al. Beta Thalassaemia in Pakistan: A Pilot Programme on Prenatal Diagnosis in Multan. *J. Pediatric Hematol Oncology* 2012;34:90-92