

## **Awareness among parents of children with thalassemia major**

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### **Abstract**

**Objective:** To determine the awareness among parents of children with thalassaemia major (TM) regarding the disease.

**Methods:** This (cross sectional) study was conducted at Paediatric OPD of Civil Hospital Karachi from May 2007 to October 2007. Parents of thalassaemic children presenting to out patient department, receiving blood transfusion from Patient Welfare Association (PWA) were interviewed using a pre designed questionnaire. Informed verbal consent was taken from the parents. Questions regarding duration of illness, awareness regarding screening of blood, mode of transmission of disease, prevention and treatment were asked.

**Results:** A total of 120 care takers were questioned. Majority was of low socioeconomic class and 66.7% were illiterate. Although nearly 100% of the patients were receiving blood transfusions either from PWA or Hussaini blood bank or both, only 15.8% knew the importance of blood screening. Knowledge regarding desferrol was present in 55% of the patient's parents but only 10.9% were receiving it adequately. Only 15% knew that thalassaemia is an inherited disorder and family screening of the siblings and antenatal diagnosis in subsequent pregnancies was done in 5.8% and 5% respectively.

**Conclusion:** Awareness of parents regarding the disease was inadequate. General public and parents of thalassaemic children should be sensitized in this regard (JPMA 58:621; 2008).

### **Introduction**

Thalassaemia, a hereditary anaemia of varying severity<sup>1</sup> is one of the commonest inherited disorders in Pakistan.<sup>2</sup> It is estimated that about 9000 children with beta thalassemia are born every year, although no documentary registry is available in Pakistan. The estimated carrier rate is 5-7%, which accounts to 9.8 million carriers in the total population.<sup>3</sup>

The cultural and religious scenario in Pakistan is such that consanguineous marriages are quite common.<sup>4,5</sup> There is no concept of premarital screening or counseling of individuals with a family history of the disease. Furthermore antenatal diagnosis is not widely available.

The concept of termination of pregnancy is an ethical and religious issue in the community.

With this state of affairs, thalassaemia is best treated conservatively with all its inherent complications eventually resulting in death, unless a definitive treatment like bone marrow transplantation is carried out, which itself is beyond the resources of a large segment of the population.<sup>6</sup>

The only way to prevent the disease and reduce the morbidity and mortality is by educating the general population.<sup>7</sup> For this reason in this present study, awareness among parents of thalassaemic patients regarding the disease was evaluated.

## Patients and Methods

This study was carried out at the out patient department (OPD) of Civil Hospital Karachi (CHK) for 6 months duration from May 2007 to October 2007.

Parents of all thalassaemic children coming to Civil Hospital Karachi for blood transfusion from Patient Welfare Association (PWA) blood bank were included in the study. PWA blood bank is a registered blood bank which provides blood and blood products to all the patients of CHK free of cost. The parents were interviewed using a predesigned questionnaire by a single interviewer after verbal informed consent.

Questions regarding the duration of illness, consanguinity of parents, extended family history of thalassaemia, source of blood, screening of blood, knowledge about the infections transmitted through blood products, prevention from these infections, mode of transmission of disease, their practices regarding iron chelation therapy and antenatal diagnosis were asked.

All the data was entered in SPSS version 10 and evaluated. Results have been presented in the form of frequencies and percentages where applicable.

## Results

A total of 120 care takers were interviewed. The mean age of patients was  $8.052 \pm 4.13$  years, with ages ranging from 2-18 years. There was a slight preponderance of females which accounted for 57.5% of the patients. Forty one percent were Balochi, 14.2% were Sindhis while 12.5% were Pathans.

One hundred ten (91.7%) had an income <Rs 5000/month. Nearly two-third (66.7%) were illiterate with only 2.5% with a higher education. Consanguinity was positive in 82.5% of the parents with extended family history of thalassaemia positive in 40.8%. Only 39.2% of the patients were registered at PWA blood bank. Sixty eight (68.3%) were receiving blood from Patients Welfare Association, 5% from Hussani while 26.7% from both. Majority (89.2%) had no idea about screening of blood. Only 7.5% knew that blood should be screened for hepatitis B,C and HIV while the remaining 3.3% believed that screening should be done for cold, fever, diarrhoea and mismatch and 5% did not have an idea for what illnesses screening should be done. Only 12.5% were immunized against Hepatitis B. Around 27.5% did not know if they should be immunized. Fifty five percent knew children should receive Desferrol (Table). Only 10.9% received it appropriately while 37.5% received it only at the time of transfusions which was given intravenously. Only 23.3% knew that it has to be given subcutaneously. Forty one percent knew that a pump is used to give Desferrol (Table).

Table: Awareness regarding Chelation Therapy.

Knowledge about Desferral	66 (55%)
<b>How often child is receiving Desferral</b>	
Daily	2 (1.7%)
Alternate days	11 (9.2%)
Monthly with blood transfusion	45 (37.5%)
Not receiving	61 (50.8%)
<b>Knowledge about how it should be given</b>	
Intravenous	11 (9.1%)
Subcutaneous	28 (23.3%)
Not known	81 (67.5%)
<b>How it is given to your child</b>	
Intravenous	42 (34.9%)
Subcutaneous	10 (8.3%)
Not given	68 (56.7%)
<b>Knowledge about infusion pump</b>	
Yes	50 (41.7%)
No	70 (58.3%)

Fifteen percent knew other that siblings could be affected, while 5.8% had the siblings screened. Twelve percent were aware of consanguinity to be a risk factor for thalassaemia with only 5% having undergone antenatal diagnosis.

## Discussion

Thalassaemias are a heterogeneous group of genetic disorders of human haemoglobin synthesis, characterized by imbalanced globin chain production which leads to ineffective erythropoiesis and anaemia.<sup>8</sup>

Beta thalassaemia occurs world wide, with a higher prevalence among Mediterranean population in the Middle East, in parts of India, Pakistan and South East Asia.<sup>9,10</sup>

The average life expectancy in Pakistan is 10 years and at present the disease load is of 90000 to 100000 patients throughout the country.<sup>3</sup>

The management of thalassaemia major in a developing country poses a major challenge to the health services. Lack of facilities and coordination to this multi disciplinary problem make the treatment difficult in a variety of ways.<sup>6</sup> Awareness about the disease among the caretakers and the availability of antenatal diagnosis is not available readily. Bone marrow transplantation, is out of reach for most of the parents because of financial constrains.<sup>6,11,12</sup>

Therefore, conservative treatment remains the choice in developing countries like Pakistan.<sup>6</sup> Parental awareness regarding various aspects of beta thalassaemia is of great importance not only for the prevention of thalassaemia major in the family, but also for the proper management of thalassaemic children.<sup>7</sup>

In the developed countries much attention has been directed to the prevention of disease by detection of thalassaemia carriers and marriage counseling. By using

this prevention programme in Sardinia,<sup>13</sup> the incidence of thalassaemia patients has decreased from 1:250 live births to 1:1000 live births. Similarly in Cyprus,<sup>14</sup> the incidence of thalassaemia major cases dropped by 96%. In the present study, consanguinity was quite high compared to the study done at Lahore showing that 56.7% of the couples were first cousins and 19.8% were relatives.<sup>15</sup> The results showed that 87.5% did not know that the marriages in the same family increased the risk of genetic transmission which is comparable (80%) with a study done at thalassaemia welfare centre Rawalpindi.<sup>7</sup>

Thalassaemic patients need life long blood transfusion for their survival, so it is very important that they should get blood from a reputable source because they are at high risk of acquiring several blood-borne viruses.<sup>17,18</sup> In the present study, almost all the patients were receiving blood from registered blood banks, but despite this fact only 15.8% knew that blood should be screened and 89.2% had no idea about the illnesses for which screening should be done. This is comparable with studies from other parts of the country showing that 80% were unaware of the importance of screening of blood.<sup>7</sup>

The prevalence of hepatitis B, C and HIV is very high in thalassaemic patients because of repeated blood transfusions. A study done at Quetta showed that 30% of thalassaemics were positive for anti HCV, 14% for hepatitis BsAg and 0.7% for anti HIV antibodies<sup>19</sup> and 8.4% and 56.8% from a study at Peshawar<sup>20</sup> and 42% seropositivity for anti HCV antibodies from other parts of the country.<sup>21</sup> Only 12.5% of patients had received hepatitis B immunization, while 27.5% were not aware of it. Reasons for not being immunized were lack of knowledge on the part of parents, improper counseling by the doctors and non affordability.

Hemosiderosis is a major cause of death in patients with thalassaemia major. So for improving their quality of life, chelation therapy is of prime importance. Iron chelation therapy with desferrioxamine in patients with thalassaemia major has dramatically altered the prognosis of this previously fatal disease.<sup>23,24</sup> Only 10.9% were receiving chelation adequately which was higher than reported from a study done at Rawalpindi,<sup>7</sup> while 37.5% received it intravenously only at the time of transfusions. Reasons for inadequate chelation were again lack of knowledge, poverty, unavailability of infusion pumps and poor compliance of the patients who were given desferol by subcutaneous route. Even though 40% of the thalassaemic children were registered with PWA, their knowledge, awareness, and chelation was poor because the only facilities provided to them, is blood transfusion in the emergency without being followed by a doctor or trained

health staff. Poor compliance to blood transfusion and chelation therapy has markedly reduced the life expectancy in our patients as compared to western population.<sup>25</sup>

In this study, only 5% had undergone antenatal screening and 5.8% got their siblings screened for thalassaemia major. The reasons for this being the lack of diagnostic facilities, family's elders influences, religious beliefs, besides the universal factors of lack of knowledge and poverty.

The present study concluded that the awareness of parents regarding this chronic illness was inadequate and these patients will continue to suffer a slow and painful course ultimately leading to death.

## References

- 1) Takeshita K. Thalassaemia, Beta [online] 2006 [cited 2006 Nov 15]. Available from: URL: <http://www.emedicine.com/med/topic2260.html>.
- 2) Khateeb B, Moatter T, Shaghil AM, Haroon S, Kakepoto GN. Genetic diversity of beta-thalassaemia mutations in Pakistani population. *J Pak Med Assoc.* 2000; 50:293-96.
- 3) Lodhi Y. Economics of thalassaemia management in Pakistan. In *Thalassaemia Awareness Week*, Ed. Ahmed S. Friends of Thalassaemia 2003.
- 4) Ahmed S, Saleem M, Modell B, Petrou M. Screening extended families for genetic hemoglobin disorders in Pakistan. *N Engl J Med.* 2002 10; 347:1162-68.
- 5) Bittles AH. The role and significance of consanguinity as a demographic variable. *Popul Dev Rev* 1994; 20:561-84.
- 6) Rahman MU, Lodhi Y. Prospects and future of conservative management of beta thalassaemia major in a developing country. *Pak J Med Sci* 2004; 20:105-12.
- 7) Hassan K, Aslam M, Ikram N. Parental knowledge and awareness in cases of thalassaemia major. *J Pak Inst Med Sci* 2002; 13: 623-26.
- 8) Kaczorowska-Ha? B, Balcerska A, Zdebska E, Wlaz?owski M. Thalassaemia beta in children from Pomerania Region *Med Wieku Rozwoj.* 2007; 11:69-72.
- 9) Khattak MF, Saleem M. Prevalence of heterozygous beta-thalassaemia in northern areas of Pakistan. *J Pak Med Assoc.* 1992; 42:32-34.
- 10) Huang SZ, Zeng FY, Ren ZR, Lu ZH, Rodgers GP, Schechter AN. et al. RNA transcripts of the beta-thalassaemia allele IVS-2-654 C-->T: a small amount of normally processed beta-globin mRNA is still produced from the mutant gene. *Br J Haematol.* 1994; 88:541-46.
- 11) Zakerinia M, Khojasteh HN, Ramzi M, Haghshenas M. Bone marrow transplantation in thalassaemia major patients using "short" anti-thymocyte globulin therapy in Shiraz, Southern Iran. *Transplant Proc.* 2005; 37:4477-81.
- 12) Lucarelli G, Galimberti M, Polchi P, Angelucci E, Baronciani D, Giardini C. et al. Marrow transplantation in patients with thalassaemia responsive to iron chelation therapy. *N Engl J Med.* 1993 16; 329:840-44.
- 13) Cao A, Rosatelli C, Galanello R, Monni G, Olla G, Cossu P. et al. The prevention of thalassaemia in Sardinia. *Clin Genet.* 1989; 36:277-85.
- 14) Buki MK, Qayum I, Siddiqui N. Prevalence and preventive measures for thalassaemia in Hazara region of NWFP Pakistan. *JAMC* 1998; 10: 28-31.
- 15) Hafeez M, Aslam M, Ali A, Rashid Y, Jafri H. Regional and ethnic distribution of beta thalassaemia mutations and effect of consanguinity in patients referred for prenatal diagnosis. *J Coll Physicians Surg Pak* 2007; 17: 144-47.
- 16) Karimi M, Nikrooz P, Kashef S, Jamalain N, Davatolhagh Z. RBC alloimmunization in blood transfusion-dependent beta-thalassaemia patients in southern Iran. *Int J Lab Hematol.* 2007; 29:321-26.
- 17) Singh H, Pradhan M, Singh RL, Phadke S, Naik SR, Aggarwal R. et al. High frequency of hepatitis B virus infection in patients with beta-thalassaemia receiving multiple transfusions. *Vox Sang.* 2003; 84:292-99.
- 18) Mollah AH, Nahar N, Siddique MD A, Anwar KS, Hassan T, Azam MD G. Common Transfusion-transmitted Infectious Agents among Thalassaemic

- Children in Bangladesh. *J HEALTH POPUL NUTR* 2003; 21:67-71.
- 19) Kapoor C, Hanif M, Iqbal M. Poly transfused thalassemic patients; prevalence of viral markers and malarial parasite. *Professional Med J* 2007; 14: 177-80.
- 20) Shah MA, Khan MT, Ulla Z, Ashfaq NY. Prevalence of Hepatitis B and Hepatitis C virus infection in multitransfused thalassemia major patients in North West Frontier province. *Pak J Med Sci* 2005; 21: 281-84.
- 21) Yonus M, Hassan K, Ikram N, Naseem L, Zaheer HA, Khan MF. Hepatitis C virus seropositivity in repeatedly transfused Thalassemia Major patients. *Int J Pathol Jun*; 2:20-23.
- 22) Ladis V, Chouliaras G, Berdousi H, Kanavakis E, Kattamis C. Longitudinal study of survival and causes of death in patients with thalassemia major in Greece. *Ann N Y Acad Sci.* 2005; 1054:445-50.
- 23) Olivieri NF, Brittenham GM. Iron-chelating therapy and the treatment of thalassemia. *Blood.* 1997 1; 89:739-61.
- 24) Brittenham GM, Nathan DG, Olivieri NF, Pippard MJ, Weatherall DJ. Deferiprone versus desferrioxamine in thalassaemia, and T2\* validation and utility. *Lancet.* 2003 11; 361:183-84.
- 25) Rai MA, Tanoli ZM, Gandapur ASK. Clinical spectrum of patients with beta thalassemia: a review of fifty four patients. *Gomal J Med Sci* 2005; 3: 55-60.
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