MEDICAL CHANNEL

Original Article

FREQUENCY OF THALASSEMIA TRAIT IN PREGNANT WOMEN

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ABSTRACT

Objective: To determine the frequency of pregnant women with thalassemia trait at Department of obstetric and gynaecology Liaquat University hospital Hyderabad.
Methodology: This is a prospective observational study of 200 pregnant women coming to Out Patient Department (OPD) and labor ward Department of Obstetrics and Gynaecology, Liaquat University Hospital (Tertiary Care Hospital) Hyderabad during study period of one year from July 2004-June 2005. The patients were selected randomly irrespective of their gestational age. These women were screened for thalassemia trait by hemoglobin electrophoresis.
Results: During one-year study 17(8.5%) out of 200 women were diagnosed with the help of hemoglobin electrophoresis to have thalassemia minor. The age of patients ranges between 15-40 years. Out of these, 10(59%) women were married with their cousins and family history was present in 4(24%) women. One woman had thalassemic child. Husbands of 3(17.6%) women were carriers of thalassemia.
Conclusion: The frequency of thalassemia minor in pregnant women was 8.5% in this study. In 17.6% cases husbands were also carriers. This would justify routine screening for thalassemia, to identify at-risk couples due to a risk as high as 25% for affected offspring.

Key Words: Thalassemia, thalassemia major, thalassemia minor, carrier status, prenatal screening, electrophoresis.

INTRODUCTION

Thalassemias are caused by some genetic defects that lead to reduced synthesis of α or β chain of hemoglobin[1].
Beta thalassemia is caused by deficient synthesis of α- chain, whereas alpha thalassemia is caused by deficiency of α- chain. The hematologic consequences of diminished synthesis of one globin chain stem not only from low intracellular hemoglobin but also from a relative excess of the unimpaired chain leading to ineffective erythropoiesis and hemolysis[1].
Thalassemia major is characterized with severe life threatening anemia during infancy and child hood. These children required chronic transfusion therapy to correct anemia and suppress ineffective erythropoiesis [2, 3, 4].
Thalassemia intermedia is either a homozygous β-thalassemia, a combined a and β-thalassemia or β-thalassemia with high level of fetal hemoglobin F. Signs and symptoms of thalalssemia intermedia are comparable to those of thalalssemia major but are of lesser magnitude[2, 3, 4].
Patients with thalassemia minor have little or no hematologic disease and are clinically

asymptomatic. There is hereditary persistent fetal hemoglobin with microcytosis (low MCV) and hypochromia out of proportion of the severity of their anemia[2, 3].

Recognition of β- thalassemia trait is important on two grounds. One differentiation from the microcytic hypochromic anemia of iron deficiency and second genetic counseling.

In Pakistan the carrier rate is 8-10%[5]. According to unofficial estimates over 4000 thalassemia children are being born in Pakistan each year and the available health care facilities are inadequate to deal with the number of sick children. It is possible to discover up to 70% of hidden carriers by simple screening for HbA_2 by electrophoresis or column chromatography techniques, which are not unduly expensive or need highly skilled equipment or staff. Due to high prevalence of this disease in Pakistan, it is important to raise public awareness and encourage people to get tested for trait and use the option of prenatal diagnosis of couples at risk of having thalassemia major off spring[6].

Programs of prospective carrier screening and genetic counseling for β- halassemia among couples planning marriages, preconception or during pregnancy are ongoing in several at risk population in the Mediterranean area[7].

A study was conducted in Israel for screening of β-thalassemia, they concluded that a preventive program among pregnant women is feasible and prevents most of the new cases of hemoglobinopathies[8].

Pregnant women with hemoglobinopathy may have an iron deficiency, which can be treated with iron supplementation as is done for other pregnant women. So iron deficiency in pregnant women affected with β- thalassemia minor shows the need for prompt iron treatment. Depending on the cause, treating iron deficiency during pregnancy might prevent anemia and adverse effects on the pregnancy[9].

It is still not widely appreciated that the thalassemias are posing an increasingly serious health burden in many parts of the world, particularly the countries of Asia. For example, the gene frequency in Indonesia is such that to treat even a proportion of patients in the future will require between 1.25 and 1.5 million units of blood each year[10].

Thalassemias are a serious health burden in many countries of the world, requiring increasing number of blood transfusions each year[10] and the only way of controlling this serious health burden is adequate education, pre marital screening and counseling of parents for pre natal diagnosis that seems to be cost effective as compared to the life long blood transfusions required by thalassemic patients and further more, effective programs for control and care of thalassemic patients should be evolved[11].

MATERIAL & METHOD

This is a prospective study of 200 pregnant women coming to out patient department (OPD) and labor ward of Department of obstetrics and gynaecology, Liaquat University Hospital (tertiary care hospital) Hyderabad during study period of one year from July 2004-June 2005. All women included in the study were registered on a proformed performa, after taking consent. The data was collected regarding their age, parity, blood relation with husband, family history of thalassemia, history of any thalassemic children. 6 ml of blood was drawn and distributed in two K2EDTA tubes. One K2EDTA sample was analyzed by using a hematology analyzer Sysmax cell counter for Hb, RBC count, MCV, MCHC, while other sample was analyzed for HbA2 and HbF after hematology analysis showed microcytic hypochromic anemia. Samples of partner of those patients who were seen to be carriers were subjected to electrophoresis after an informed consent

Research and Diagnostic Lab, LUH was selected for hemoglobin electrophoresis. Alkaline cellulose acetate method was used. Apparatus was automated electrophoresis system by Genio Inter Lab Italy.

All data was entered in computer and mean, frequencies and standard deviation were calculated by using SPSS.

RESULTS

Out of 200 pregnant women screened, 17(8.5 %) were diagnosed as carrier of thalassemia (thalassemia trait). 01 case of thalassemia intermedia and 02 cases of sickle cell trait (table I). Mean maternal age was 27.2 ± 5.1 with youngest being 15 year and oldest being 40 year of age.

All women with thalassemia trait were anemic clinically with Hb% ranging from 3.2 gm/dl to 10 gm/dl. Family history of thalassemia was present in 4(24%) of the carrier women, rest of them were unaware of the disease or their carrier status. 10(59%) women were having cousin marriage and among 17 carrier women, 3(17.6%) were having carrier husband. One (5.89%) woman was having thalassemic child.

Red blood cell (RBC) count was low in 8(47%) cases with mean value of 3.69 ± 1.0. Mean Corpuscular Volume (MCV) was reduced in 14(82.3%) cases with mean value of 69.61 ± 3.2, Mean Corpuscular hemoglobin (MCH) was reduced in all cases with mean value 20.2 ± 3.2, and Mean Corpuscular Hemoglobin Concentration (MCHC) was reduced in 16(94%) cases with mean value of 28.2 ± 2.2. Hb A2 was raised in 14(82.3 %) of women with mean value of 4.55 ± 1.4. Hb F was raised in 11(64.7%) of women with mean value of 2.75 ± 2.

TABLE I

FREQUENCY OF THALASSEMIA TRAIT AND OTHER HAEMOGLOBINOPATHIES IN PREGNANT WOMEN

Type of haemoglobinopathy	Number	%
Thalassemia trait	17	8.5
Thalassemia intermedia	01	0.5
Sickle cell trait	03	1.5

TABLE II

FAMILY HISTORY OF THALASSEMIA AND

BLOOD RELATION OF CARRIER WOMEN WITH THEIR HUSBANDS

n=17

	Frequency	Percentage (%)
Family History	4	24
Relation With Husband	10	59

TABLE III

HISTORY OF THALASSEMIA MAJOR CHILDREN

ANY THALASSEMIC CHILD	FREQUENCY	PERCENTAGE (%)
Yes	01	5.89
No	16	94.11
Total	**17**	**100**

DISCUSSION

In Pakistan the carrier rate in general population is 8-10%[5], while our study on pregnant women shows 7.5% carrier rate. These women are likely to transmit disease, which further multiplies by social customs of cousin marriages as shown in a study conducted at Lahore where 76.6% of the couples were having cousins marriages[12] and 85% females in our study also had cousin marriages. Out of the 17 carrier women, 17.6% women were having their husband carriers of thalassemia and only one couple was having a child suffering from thalassemia major. Two out of three women with carrier husbands were in gestational age of 34 and 37 weeks, so they were counsel for the risk of disease in their offspring in the next pregnancy. While only one woman was diagnosed at the gestational age of 24 weeks. She was informed, counseled and referred for CVS sampling.

Generally the carrier patients have mild or absent anemia[13, 14]. In our study the results are somewhat different i.e all the women with thalassemia minor are anemic with Hb ranging from 3.2 to 10.8gm/dl. The low Hb levels are most probably due to presence of iron deficiency anemia being highly prevalent in our population and especially in women of low socioeconomic group and most of the women coming to LUH belong to this class.

It has been observed in different studies that the sensitivity and specificity of red cell count and red cell indices is very high for screening of thalassemia trait[13, 15]. The red cell count is elevated, while it is decreased in our study with mean value of 3.69 +/-1.08 (mean+/-SD). The MCV & MCH values are reduced in thalassemia trait. The MCHC is normal or only slightly reduced[14] our study also shows the reduced values of MCV and MCH and along with them values of MCHC are also low, mean MCV value in our study is 69.61+/-2 fl (mean+/-SD), MCH value 20.2+/-3.2pg and MCHC is 28.2+/-2.23gm/dl. Again this could be because of concomitant iron deficiency anemia.

The most consistent feature of thalassemia minor is an increase in HbA_2. Its values vary according to laboratories. The cut off value in the Diagnostic and Research Laboratory Liaquat University of Medical and Health Science is 1.5-3.5%. Hemoglobin electrophoresis reveals an increase in HbA_2 from 4% to 8% of the total hemoglobin (normal 2.5% ± 0.3%). HbF can be normal or slightly increased[1]. Raised level of HbA2 confirms significant number of patients with thalassemia trait[13]. Mean Hb A2 in our study was 4.55 +/-1.45 % (mean +/-SD). Hb F is increased slightly (1-2%) in approximately 50% of the cases[13] while in our study the Hb F was raised in 64.7% of the cases with mean value of 2.75+/-2.65(mean+/-SD).

CONCLUSIONS

The frequency of thalassemia minor in pregnant women was 8.5% in this study. In 17.6% cases husbands were also carriers. This would justify routine screening for thalassemia, to identify at-risk couples due to a risk as high as 25% for significantly affected offspring. Public awareness programs must be launched to inform people regarding prevalence of the genetic disorders, especially thalassemia. Each individual must know his/her blood group as well as thalassemia status.

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