Section: Pathology

Original Research Article

# A STUDY OF THE PREVALENCE OF THALASSEMIA AND IT'S CORRELATION WITH LIVER FUNCTION TEST IN DIFFERENT AGE AND SEX

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## Abstract

**Background:** Thalassemia is the most common genetic blood disease in the world and varies in different population group in the world. World Health Organization (WHO) estimates that at least 6.5% of the world populations are carries of different inherited disorders of hemoglobin.[2] **Materials and Methods:** The study was designed on the basis of prospective observational type of study.S ubject collection and study procedure Subjects were collected on every working day of the week from 10:00 am to 2:00 pm from the Department of Thalassaemia and when needed from the Biochemistry Laboratory in the, Govt. Medical College, Purnea, Bihar. **Result:** Clinico-hematological study of thalassemia was done on 50 patients. Salient features observed in this study were: Age Mean (±SD) age in total 50 patients between the age group 2 and 10 years was 4.666 ± 3.91617, between 11 and 20 years mean was 18.1575 ± 2.10208 and between age 21 and 30 years mean was 22.1035 ± 2.74510 were include in the study. **Conclusion:** overall study, we tried to correlate the liver damage in thalassemic patients with a hope to improve the liver damage.

## INTRODUCTION

Thalassemia is the most common genetic blood disease in the world and varies in different population group in the world.[1] World Health Organization (WHO) estimates that at least 6.5% of the world populations are carries of different inherited disorders of hemoglobin.[2] Another WHO report estimates that 3% are carriers of beta-Thalassemia and 4% are carriers of Hb E in India. In India, more than 7000 children are born with Thalassemia each year.[3] Majority are born in countries with limited resources where priority tends to be given to tackling high rates of infant and child mortality from infection diseases and malnutrition.[4] The patients suffering from beta-thalassemia major and Hb E/ beta-thalassemia do not survive for more than 5 years without blood transfusion.[5] Thalassemia (also known as Mediterranean anemia, Cooley's anemia, Beta-thalassemia or Alpha-thalassemia) is an inherited blood disorder affected by an abnormal form of hemoglobin blood disorder is the most common inherited single gene disorder in the world [Figure 1]. This specific type of blood disease results in excessive destruction of red blood cells which in turn leads to anemia.[6] For a better understanding of this disease one must know the importance of hemoglobin.[7] In vertebrates, hemoglobin is the iron-containing oxygen-transport protein that is found in red blood cells which carries oxygen from the lungs to the rest of the body and then brings the carbon dioxide back to the lungs to be dispensed. People who have thalassemia produce fewer healthy hemoglobin proteins, and their bone marrow produces fewer healthy red blood cells. With too few normal red blood cells, not enough hemoglobin is available to help carry oxygen to the body.[8] Thalassemia occurs more often among certain ethnicities, including people of Italian, Greek, Middle-eastern, Asian and African descent. Thalassemia is an inherited disorder which means they are passed from parents to their children.[8] a- thalassemia occurs commonly in the people of those from Southeast Asia, the Middle-east, China and those of African-American descent.[9] b-thalassemia occurs commonly in people of the Mediterranean region, Chinese, other Asians and African-Americans.[10] This study revealed that the overall prevalence of beta-thalassemia trait in India was 4.1% and Hb E trait 6.3%.[11] A recent study showed that carrier status of Hb-E is 6.1% and as high as 40% in tribal children in India.[12]

## MATERIALS AND METHODS

The study was designed on the basis of prospective observational type of study. In the study the current


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data was obtaining form a specific diseased group with a progressive complication. Department Of Pathology, Govt. Medical College, Purnea, Bihar.

Subject collection and study procedure Subjects were collected on every working day of the week from 10:00 am to 2:00 pm from the Department of Thalassaemia and when needed from the Biochemistry Laboratory in the, Govt. Medical College, Purnea, Bihar. Parents or guardians were informed of the purpose of the study. For each patient a detailed history was taken from mother or the attendant. After taking brief history preliminary selection was done, and the purpose to the study was explained in details to its subject. After taking consent from the parents, data was collected, which included sex, age at presentation, age at diagnosis and clinical symptoms at presentation. A thorough physical examination was done in each patient. Majority of the patients diagnoses were confirmed by Hb-electrophoresis. The patients who fulfilled the inclusion criteria were included for this study. All the planned information obtained and recorded in the data collecting sheet properly. A total of 50 subjects were included in this study. There was no specific preference for race, religion and socioeconomic status.

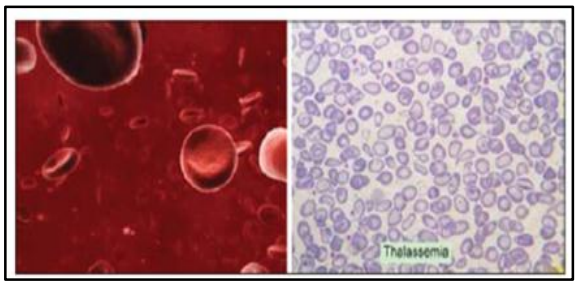


**Figure 1: Blood film showing characteristics normal red blood cell and abnormal pale red blood cells**

**Inclusion Criteria:**

Patients with confirmed diagnosis of thalassemia were randomly selected as first as diagnosis up to the age 30.[13] The subjects on regular blood transfusions were enrolled. The inclusion criteria considered in data collection are: (1) Thalassemia major, (2) History of jaundice other than viral, alcohol or heavy metal induces jaundice, (3) Repeated blood transfusion, (4) History of taking iron chelating therapy and (5) Clinically diagnosed Hepatomegaly.

**Exclusion Criteria:**

There is more problem to exclude subject only basis on clinical questionnaire and on the basis of some laboratory test.[14] The exclusion criteria are as follows: (1) Thalassemia trait or intermedia type, (2) History of jaundice due to viral, alcoholic or heavy metal induces, (3) History of spleenectomy, (4) Age more than 30 years.

Additional investigations done were: • Observation of liver function test (LFT; Serum billirubin) in different thalassemic patients. • Estimation of serum billirubin (Jendrassik and Grof method) by spectrophotometric/ filter photometric test.

**Statistical analysis:** Statistical analysis was carried out using SPSS statistical package (version 11.5). Analysis of variance (ANOVA) of the data was used to detect overall difference in group means. Differences among group means were assessed using least significance difference (LSD).

## RESULTS

Clinico-hematological study of thalassemia was done on 50 patients. Salient features observed in this study were: Age Mean (±SD) age in total 50 patients between the age group 2 and 10 years was 4.666 ± 3.91617, between 11 and 20 years mean was 18.1575 ± 2.10208 and between age 21 and 30 years mean was 22.1035 ± 2.74510 were include in the study.

**Sex:** Mean (±SD) of sex in 50 no. of patients in male sex 13.000 ± 9.76147 and females are 14.8524 ± 6.00821

**Type of thalassaemia:** of the type of thalassemia E-beta-thalassemia 40 in no. (77.4%), beta-thalassemia 7 in no. (15.1%) and beta major 3 in no. (7.5%).

**Total serum billirubin (TSB, mg/dl):** Mean (±SD) TSB in total 50 patients with age 2-10 years and 11-20 years are 3.056 ± 1.81673 and 4.7177 ± 1.23664, respectively. Statistically no significant billirubin difference was observed between these two groups. P > 0.363

Mean (±SD) TSB in total 50 patients with age group 11-20 years and age 21-30 years are 4.7168 ± 1.35663 and 6.2626 ± 1.01010, respectively. Statistically no significant billirubin difference was observed between these two groups. P > 0.091

Mean (±SD) TSB in total 50 patients with age group 21-30 years and 2-10 years are 6.2626 ± 1.01011 and 3.6045 ± 1.8153, respectively. Statistically significant billirubin difference was observed between these two groups. P < 0.001 Mean (±SD) TSB in total 50 patients with different sex group are 5.1613 + 1.24137 and 4.7077 ± 1.20171 for female and male, respectively. Statistically no significant billirubin difference was observed between these two groups. P > 0.491

**Result from SPSS:** From the data it is easy to interpret that, there is a significant difference found in S. Billirubin with age group 2 to 10 years and 21 to 30 years of age.

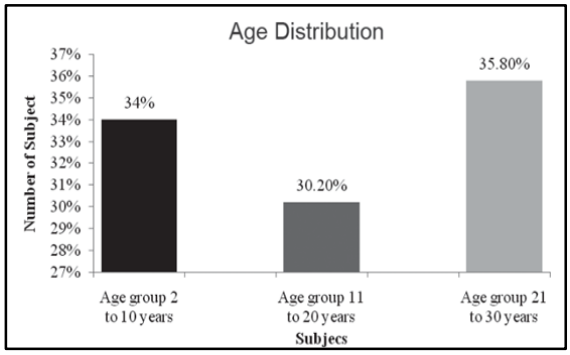


**Figure 2: Colum diagram showing age distribution over LFT**


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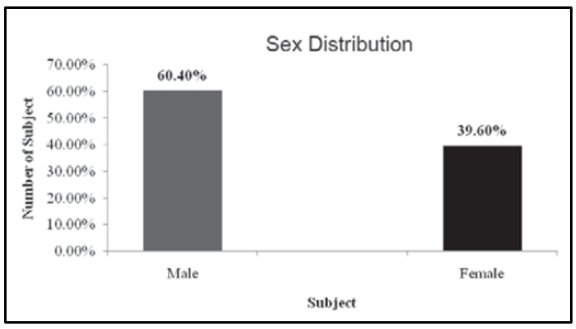


**Figure 3: Colum diagram showing sex distribution over LFT**

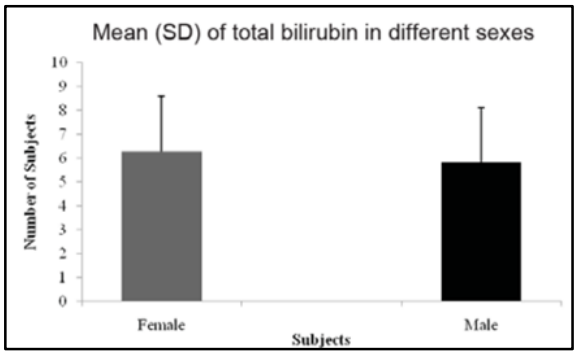


**Figure 4: Colum diagram show mean (±SD) of total billirubin (mg/dl) in different sexes**

## DISCUSSION

Thalassemia a group of genetic disorder occur mainly due to defective formation of globin chain of the hemoglobin moiety of the RBC. This specific type of blood disease results in excessive destruction of red blood cells which in turn leads to anemia. In this disease RBC breakdown occur at an early stage due to abnormal globin chain unable to protect RBC in oxidative stress. Resulting destruction of RBC leads to produce billirubin production which ultimately metabolized in liver for excretion. In thalassemia the rate of destruction of RBC is so rapid that it exceeds the liver capacity to metabolize the excess billirubin.[16-18] Hb E is the most common variant hemoglobin with a mutation in beta-globin gene causing substitution of glutamic acid for lysine at position 26 in beta-globin chain. Hb E Disease presents in 3 forms namely heterozygous state (Genotype AE or Hb E trait), homozygous state (Genotype EE or Hb E disease) and compound heterozygous states [1. HbE beta-thalassemia (E-beta-thalassemia) 2. Sickle Cell/Hb E Disease (SE Genotype)].[19-21] Pathophysiology is complex which involves ineffective erythropoiesis, apoptosis, Oxidative damage and shortened red cell survival. Interaction between Hb E and betathalassemia alleles is main determinant in pathophysiology. Hb F level is strongest predictor of morbidity. Hb E Trait may □□0-thalassemia or □be coinherited with either -thalassemia. The compound heterozygous state is quite common in Thailand and occurs throughout a large part of Southeast Asia stretching from Indonesia to Sri Lanka, Northeast India andBangladesh with prevalence rate of 30-40%, with very few. Pediatric cases being reported from India.[22-24] In the study I enrolled LFT to observe the liver damage in thalassemic patient. I focus mainly the basic LFT such asS. Billirubin. S. billirubin is the first point that helps to recognized jaundice patient. I tried to find out the actual liver damage (hepatocellular/obstructive) in thalassaemic patient in different age group and different sex. In accordance with other studies the most common recognized abnormality was excess billirubin turnover with excess RBC damage in relation to liver damage (change in ALT level with age). Three principal clinical pictures have been recognized: a. acute intrahepatic cholestasis, b. hepatic crisis and c. lithiasis. (Curcio et al., BMC Gastroenterology 2010, 10:117). Cholelithiasis and choledocholithiasis are common complications in patients with sickle cell disease or thalassemia.[25] The altered shape of red blood cells favors intravascular hemolysis and thus occlusion of the liver vascular bed, leading ultimately to tissue injury. Furthermore, hemolysis induces deposition of billirubin causing intrahepatic cholestasis and cholelithiasis.[26]

## CONCLUSION

From the overall study, we tried to correlate the liver damage in thalassemic patients with a hope to improve the liver damage in such patient with the goal of: (A) To prevent liver disease caused by viral hepatitis, iron overload, drug toxicity or hepatocellular carcinoma and (B) To monitor liver abnormalities routinely, and provide treatment for iron overload and any underlying liver disorder. The study revealed that in thalassemia patients when the age increases, the disease is progress with their complication. And in case of hepatic complication it is mainly due to hepatocellular in nature than that of obstructive one.

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