Original Article

Clinico biochemical abnormalities in Thalassemia

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Abstract

Problem statement: Thalassemias are a group of inherited disorders of Hb synthesis characterised by a reduced rate of production of one or more globin chains of Hb resulting in an anaemic state. Blood transfusion which both elevates the anaemia and suppresses the compensatory mechanism is the basis of therapy. But the inevitable consequence of prolonged transfusion therapy is the hemosiderosis. This causes complications like endocrine abnormalities along with hepatic and myocardial siderosis. The group with regular blood transfusion has the more chance of iron excess. The patients with inadequate transfusion are not free from the danger of iron overload because in them the anaemia causes excessive iron absorption. The endocrine glands are not also free from the burnt of hemosiderosis The excess iron in the storage form of ferritin get deposited in endocrine organs like pituitary, pancreas, parathyroid, and thyroid. **Methods:** It was performed on selected patients with diagnosis of Thalassemia disease, who were attending in both institution (Malda Medical College and M.G.M Medical College and L.S.K Hospital,)The 30 patients were included in our study. In the present study maximum number of cases were in the age group of 5-15 years (68%) and 32% of patients were in the age group of 16-25 years. **Results:** Dysfunction of the thyroid gland is less frequent than other endocrinopathies in thalassemia patients. Results are Distribution of Cases in Different Types of Thalassemia: Growth Hormone Study, Gonadotrophins Study. In our study no patient had yet developed hypothyroidism but possibilities of hypothyroidism in future specially of the sub-clinical hypothyroid patients remain.

Keywords: Thalassemia, hemosiderosis, Gonadotrophins, hypothyroidism.

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INTRODUCTION

Thalassemia patients are usually symptomatic during second 6 months of life as a progressively haemolytic anaemia. Failure to thrive and gain weight normally, progressive pallor, feeding problem, progressive enlargement of abdomen due to splenomegally are the common presenting symptoms. Regular blood transfusion becomes necessary. Spleen and liver enlarged due to extramedullary erythropoesis and hemosiderosis. Regular transfusion regime help the child to grow normally over the next decade. Growth retardation is more marked as the child approaches puberty. Puberty may be delayed or absent because of secondary endocrine abnormalities, anaemia and sometimes due to side effects of iron chelator. The nature of a so common disorder was not known before 1925. The historical development of knowledge of thalassemia can be categorised into 3 stages : 1)1925-1940 – the first description of different types of homozygous and heterozygous thalassemia was given by many respectable persons. 2) In the period between 1940-1950 – the genetic basis of this disorder was recognised. 3) Since 1950 onwards – different workers worked separately to find the biochemical nature of thalassemia and the distribution of the diseases. Gradually it was found out that, that is not only localised in Mediterranean region but widely distributed throughout the world. The term thalassemia was first used by Whipple and Bradford in 1932 in their classic paper as pathology of the conditions. This is a Greek word which means ‘‘Sea’’[1].

Thalassemia in India: Chatterjee and his colleagues[2] in Calcutta showed that the β thalassemia genes were common in various Indian population. It was also common in American Negros in 1950s. £ and β thalassemia were also found in African origin people

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throughout the work. Thalassemia is now known to be a disease of single Gene disorder causing a major public health problem in World population. (Weatherall and Clegg 1980)[3]. About 3% of world's population (150 million) carry β thalassemia gene. The highest prevalcea of carrier state focussed in descending order - Sardinia (11- 34%), Cicilia (10%), Greece (5- 15%). Haemoglobin is the oxygen carryings pigment of RBC in vertebrates. It is a globular molecule (Mol. Weight 64450) made up of 4 subunits. Each subunit contains a heme moiety conjugated to a polypeptide chain. Heme is an iron containing porphyrin derivative. The polypeptides are collectively referred to as globin portion of the haemoglobin molecule. There are 2 pairs of polypeptides in each Hb molecule. In normal adult haemoglobin (HbA), the two types of polypeptides are called the £ chains and the β chains. Each £ chain contains 141 amino acids and each β chain contains 146 amino acid residues. Thus HbA is designated as $£_2$ $β_2$.[4]. HbF contains Y polypeptide chains in place of the B chains of HbA and can be represented as $α_2γ_2$. Its resistance to denaturation by strong alkali is usually used in its quantitation. After the 8th gestational week HbF is the predominant haemoglobin,

MATERIALS AND METHODS

Study Place: It was performed on selected patients (outdoor) with diagnosis of Thalassemia disease, who were attending in this institution (Malda and M.G.M Medical College and L.S.K Hospital,)

Study Design: A prospective, descriptive clinical study.

Inclusion Criteria

- Patients are in the age group of 5-25 years.
- Patients are diagnosed to be suffering from β thalassemia major or Eβ thalassemia with Hepatosplenomegaly and Pallor.

Biochemical Investigation

Blood were collected from each individual by vein-puncture and were allowed to clot at room temperature for one hour. Clotted blood was centrifuged at 3000 rpm for 15 minutes and separated serum to be used for biochemical tests- **T_3, T_4, TSH, LH and FSH** and estimation of basal **GH level** in serum, clotted blood is drawn in the morning.

Statistical Analysis

Comparison of changes were made with the help of SPSS, t-test was calculated on the basis of mean and standard error of mean. Significance was considered at the level of **$p<0.02$**. Comparison was made between normal control with other group.

RESULTS

The study it reveals that out of 30 patients 10 patients had been suffering from Homozygous β thalassemia and 20 patients from E- β thalassemia. So incidence of E- β thalassemia are distinctly more than Homozygous β thalassemia. This increased incidence of E- β thalassemia indicates that these patients often suffer from anaemia requiring repeated blood transfusion and expose them to the chance of increased ferritin level in blood and haemosiderosis like β thalassemia major. Among the age group there is no patients in 5-10 years group. This is due to the fact tht high serum ferritin level is uncommon in this age group. In the age group of 10-15 years there are 20 patients (68%) and 10 patients are in the age group of 16-25 years. Though chances of Haemosiderosis increase with age, more patients are in the younger age group and this is due to the fact that some patients die of various complications of thalassemia before attending the elder age. In this study 30 patients belong to 30 families. Of these 2 families have 1 sibbling each, 10 families have 2 sibblings each and 13 families have >2 sibbling each. So most of the families have more than 2 sibblings. This may be due to the fact that the parents had not adopted family planning to have children without the disease along with their child suffering from thalassemia. In our study of 30 patients Basal G.H. level is below 1 ng/ml in 2 patients i.e., in 6% of patients and G.H. level is above 1 ng/ml in 94% of patients. Laboratory reference of G.H. level is 0-2 ng/ml and hence it can be said that this 5% of patients is in the lower range of G.H. level. Other 28 patients (95%) have no abnormalities in their Basal G.H. level. 30 patients, 28 patients have G.H. level above 10 ng/ml and 2 patients (6%) have G.H. level below 10 ng/ml. So these 2 patients have obviously G.H. deficiency as well as deficiency of G.H. reserve. Out of 13 female patients 31% patients had decreased and 69 % patients had normal FSH value. And 16 male patients 48 % patients had decreased and 52% patients had normal LH value. 13 female patients 30.7% patients had decreased and 69.2% patients had normal LH value. 12 female patients 36% patients had decreased and 64% patients had normal LH value. all 30 patients had T_3 and T_4 level within normal range and 6 patients (18%) had raised TSH level.

CONCLUSION

Hormonal changes of some of the common hormones namely Growth hormone (Fasting and after Insulin induced hypoglycemia),LH, FSH. TSH, T_3 andT_4 in thalassemia patients with increased serum ferritin level. We know that functions of the hormones like GH, LH and FSH, T_3, T_4, TSH all have important functions in the body and deficiency of them increase the morbidity and the mortality of the thalassemic patients inspite of

adequate blood transfusion. So to give these patients normal or near normal life the matter of endocrine abnormalities must be kept in mind along with other modalities of treatment. For this they need use of iron chelators and the use of different hormones as and when necessary.

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