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Assessment of haematological parameters in thalassemia in paediatric patients

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Abstract
Thalassemia is a blood disorder passed down through families (inherited) in which the body makes an abnormal form of haemoglobin. Haemoglobin is the protein in red blood cells that carries oxygen. The disorder results in large numbers of red blood cells being destroyed, which leads to anaemia.

The study was conducted in the Department of Paediatrics in from Sri Krishna Medical College and Hospital. The age of the patients is ranging from 1 year to 15 years. The data from the 100 patients were collected and presented as below. These patients had been receiving blood transfusions regularly.

The Thalassemia patients are having lower Haemoglobin concentration as compared to normal patients. The Hematocrit level in the Thalassemia patients is lower as compared to normal patients. The Red to White Cell distribution and mean Corpus Volume are seen higher in Thalassemia patients. The levels of the RBC are seen lower and levels of the WBC is seen slightly higher side.

Keywords: thalassemia, haemoglobin, haematological profile etc.

Introduction

Thalassemia is a blood disorder passed down through families (inherited) in which the body makes an abnormal form of haemoglobin. Haemoglobin is the protein in red blood cells that carries oxygen. The disorder results in large numbers of red blood cells being destroyed, which leads to anaemia.

Thalassemias are inherited blood disorders characterized by abnormal hemoglobin production. Symptoms depend on the type and can vary from none to severe. Often there is mild to severe anemia (low red blood cells). Anemia can result in feeling tired and pale skin. There may also be bone problems, an enlarged spleen, yellowish skin, dark urine, and among children slow growth [1].

Thalassemias are genetic disorders inherited from a person's parents. There are two main types, alpha thalassemia and beta thalassemia. The severity of alpha and beta thalassemia depends on how many of the four genes for alpha globin or two genes for beta globin are missing. Diagnosis is typically by blood tests including a complete blood count, special hemoglobin tests, and genetic tests. Diagnosis may occur before birth through prenatal testing [2].

Treatment depends on the type and severity. Treatment for those with more severe disease often includes regular blood transfusions, iron chelation, and folic acid. Iron chelation may be done with deferoxamine or deferasirox. Occasionally, a bone marrow transplant may be an option. Complications may include iron overload from the transfusions with resulting heart or liver disease, infections, and osteoporosis. If the spleen becomes overly enlarged, surgical removal may be required [3].

As of 2013, thalassemia occurs in about 280 million people, with about 439,000 having severe disease. It is most common among people of Italian, Greek, Middle Eastern, South Asian, and African descent. Males and females have similar rates of disease. It resulted in 16,800 deaths in 2015, down from 36,000 deaths in 1990. Those who have minor degrees of thalassemia, similar to those with sickle-cell trait, have some protection against malaria, explaining why they are more common in regions of the world where malaria exists [4].

Both α- and β-thalassemias are often inherited in an autosomal recessive manner. Cases of dominantly inherited α- and β-thalassemias have been reported, the first of which was in an Irish family with two deletions of 4 and 11 bp in exon 3 interrupted by an insertion of 5 bp in the β-globin gene. For the autosomal recessive forms of the disease, both parents must be carriers for a child to be affected. If both parents carry a hemoglobinopathy trait, the risk is 25% for each pregnancy for an affected child.

Estimates suggest that approximately 1.5% of the global population (80 - 90 million people) are β-thalassemia carriers. However, exact data on carrier rates in many populations are lacking, particularly in developing areas of the world known or expected to be heavily affected. Because of the prevalence of the disease in countries with little knowledge of thalassemia, access to proper treatment and diagnosis can be difficult. While there are some diagnostic and treatment facilities in developing countries, in most cases these are not provided by government services, and are available only to patients that can afford them. In general, poorer populations only have access to limited diagnostic facilities together with blood transfusions. In some developing countries, there are virtually no facilities for diagnosis or management of thalassemia [5].

Normally, the majority of adult hemoglobin (HbA) is composed of four protein chains, two α and two β globin chains arranged into a heterotetramer. In thalassemia, patients have defects in either the α or β globin chain, causing production of abnormal red blood cells (In sickle-cell disease,

the mutation is specific to β globin).

The thalassemias are classified according to which chain of the hemoglobin molecule is affected. In α-thalassemias, production of the α globin chain is affected, while in β-thalassemia, production of the β globin chain is affected.

The β globin chains are encoded by a single gene on chromosome 11; α globin chains are encoded by two closely linked genes on chromosome 16 [6]. Thus, in a normal person with two copies of each chromosome, two loci encode the β chain, and four loci encode the α chain.

Hence the study has been planned to know the haematological parameters in children' suffered from the Thalassemia.

Methodology

The study was conducted in the Department of Paediatrics in from Sri Krishna Medical College and Hospital. The age of the patients is ranging from 1 year to 15 years. The data from the 100 patients were collected and presented as below. These patients had been receiving blood transfusions regularly. The approval of the institutional ethic committee had been taken before the study. All the patients were informed consent. The aim and the objective of the study is conveyed to all patients.

Results & Discussion

In the current planned study the data from 100 patients were collected and presented as below. The following is the blood group distribution in the selected patients. There are more patients of Blood group O and B are seen in the study group.

Table 1: Blood group distribution

Blood Group	No. of Patients
O	30
A	12
B	40
AB	18
Total	100

Table 2: Observation of Thalassemia

Group	Haemoglobin Concentration
Thalassemia Patients	6.8 – 7.4 gm%
Normal Patients	11.2 – 13.5 gm%

Table 3: Haematological Parameters

Group	Hematocrit Level	RDW.CV	RBC	WBC
Thalassemia Patients	20.5- 21.8 %	21.2 – 22.1%	2.2-2.9 x 10^6cmm	11.8-12.5 x 10^3 cells
Normal Patients	37 – 47 %	11 – 14 %	4.5-6.5 x 10^6cmm	4.5 -11.0 x 10^3 cells

The Thalassemia patients are having lower Haemoglobin concentration as compared to normal patients. The Hematocrit level in the Thalassemia patients is lower as compared to normal patients. The Red to White Cell distribution and mean Corpus Volume are seen higher in Thalassemia patients. The levels of the RBC are seen lower and levels of the WBC is seen slightly higher side.

Thalassemia is a worldwide disorder. α- and β -thalassemia are the most common single-genehaemoglobin disorders in the world. It is more prevalent in areas endemic for malaria [18]. South East Asia, [7-9] India, [10] Mediterranean region [11] and Middle East including Saudi Arabia [12] are the regions from where large number of cases are reported. The change in haematological parameters depends on the type of thalassemia. The clue for thalassemia is low mean corpuscular volume (MCV) < 78 fl or low meancorpuscular haemoglobin (MCH) <27 pg. Although iron deficiency is the most common cause of a low MCV ora low MCH, it is likely that this finding will point to thalassemia in regions of countries with thalassemia-proneethnic populations. There are several causes of the anaemia produced by different abnormal haemoglobins. Microcytic hypochromic anaemia is a common haematological abnormality in clinical practice and usually is caused by iron deficiency and thalassemia trait.

Conclusion

Nationwide Government sponsored programme can effectively reduce the occurrence of new cases of serious haemoglobin variants as well as thalassaemia major cases and thus making it possible to direct the available resources towards the optimization of treatment of the patients who are already present. Detection of these patients with abnormal haemoglobins will help in prevention of more serious Hb variant cases.

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