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Thalassemia: A Case Study.

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ABSTRACT

Thalassemia is an genetic blood disorders inherited from a person's parents. The two main types are alpha thalassemia and beta thalassemia. The disorder results in excessive destruction of red blood cells which leads to anaemia. The symptoms include bone deformities, delayed growth and development, excessive tiredness and fatigue, enlarged spleen. Complete blood count, haemoglobin electrophoresis and DNA testing is the diagnostic evaluation. The treatment includes blood transfusion, Iron chelation and folic acid bone marrow transplant is definitive cure available for thalassemia patient. Genetic counselling is the only prevention

Keywords: Thalassemia, electrophoresis, iron chelation bone marrow transplant, genetic counselling

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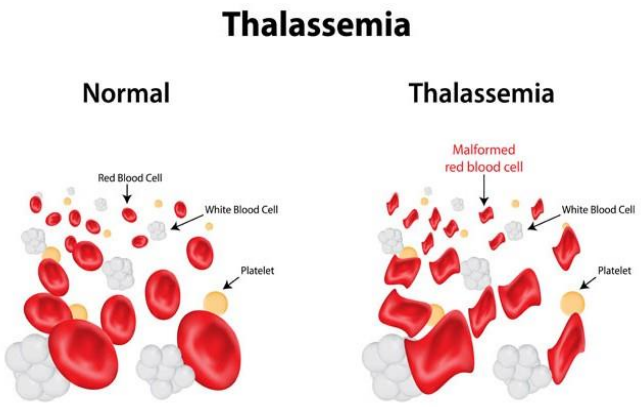
INTRODUCTION

The thalassemia is inherited disorders of hemoglobin (Hb) synthesis. Their clinical severity widely varies, ranging from asymptomatic forms to severe even fatal entities.

In most patients with thalassemia traits, no unusual signs or symptoms are encountered. Some patients, especially those with somewhat more severe forms of the disease, manifest some pallor and slight icteric discoloration of the sclera with splenomegaly, leading to slight enlargement of the abdomen. An affected child's parents or caregivers may report these symptoms. However, some rare types of β thalassemia trait are caused by a unique mutation, resulting in truncated or elongated β chains, which combine abnormally with α chains, producing insoluble dimers or tetramers. The outcome of such insoluble products is a severe hemolytic process that needs to be managed like thalassemia intermedia or, in some cases, thalassemia major. [1-5]



CASE STUDY OF BABY X

Baby x 10 years old female presented with history of high temperature and puffiness of face. She was admitted for blood transfusion. Baby x past history, the child was normal till 6 months. After which the parents noticed yellowish decolourisation of the skin, eye and fever for one week at 8 month and was treated for the same and was diagnosed as thalassemia. There after the child was on blood transfusion 15 days once and after 4 years the child undergoes every one month. The child has undergone spleenectomy surgery at 5 years of age. Baby x belongs to nuclear family there is no history of communicable disease and also known hereditary diseases. Parents are not consanguineous marriage. The children in the family are normal

Findings on 1st presentation:

- Mild thalassemic features.
- Severe growth retardation (below 3rd centile).
- Liver: 8cm.
- Spleen: 14 cm.
- Hb: 6.8 g/dl.
- S. Ferritin: 8600 ug/l

CAUSES AND RISK FACTORS

Thalassemia occurs when there is an abnormality or mutation in one of the genes involved in hemoglobin production. This genetic defect is inherited from the parents. If only one of the parents is a carrier for thalassemia, then the child may develop “thalassemia minor.” If this occurs, the child will probably not have symptoms, but will be a carrier of the disease. Some people with thalassemia minor do develop minor symptoms. If both parents are carriers of thalassemia, 25 percent chance of inheriting a more serious form of the disease.

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Thalassemia is most common in people from Southeast and Central Asia, the Mediterranean, the Middle East, India, and North Africa.

PATHOPHYSIOLOGY

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 thalassemia are classified according to which chain of the hemoglobin molecule is affected. In α-thalassemia, 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. Thus, in a normal person with two copies of each chromosome, two loci encode the β chain, and four loci encode the α chain. Deletion of one of the α loci has a high prevalence in people of African or Asian descent, making them more likely to develop α-thalassemia. β-Thalassemia.

CLINICAL MANIFESTATION

BOOK PICTURE	PATIENT PICTURE
FUSSINESS	PRESENT
PALENESS	PRESENT
FREQUENT INFECTION	PRESENT
POOR APPETITE	PRESENT
FAILURE TO THRIVE	-
JAUNDICE (YELLOWISH OF THE SKIN AND WHITES OF THE EYES	PRESENT
ENLARGED ORGANS	SPLEENCTOMY
ANAEMIA	PAIN IN JOINTS AND MUSCLES

DIAGNOSTIC EVALUATION

BOOK PICTURE	PATIENT PICTURE
COMPLETE BLOOD COUNT	The patient reveals anaemic, red blood cells are few in number
PERIPEHERAL SMEAR	Hypochromia anisocytosis poikilocytosis,microcytosis,nucleated RBC
SERUM BILIRUBIN	Moderately elevated
SERUM IRON	Elevated
BONE MARROW STUDY	Hyper cellular and erythroid hyperplasia
OSMOTIC FRAGILITY	Decreased fragility

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MANAGEMENT

MEDICAL MANAGEMENT	MANAGEMENT FOR BABY X
Iron and folic acid supplement	**Folic acid (pterylglutamic acid) 5mg/oral/bd**
Diuretics	frusemide 20 mg/oral/bd
Iron chelation therapy	**defarasiron 500mg/oral/od**
Blood transfusion	**transfusion of blood according to weight of the child**

PATIENT EDUCATION

- Genetic counselling provide detailed instruction
- Prevention & treatment of infection
- Medication
- Dietary modification to limit iron intake
- Signs of complications

REFERENCES

[1] Abraham. Paediatrics. 1st ed. Singapore: Mc. Graw Hill international company; 2001.
[2] Beharmann. Text Book of Paediatrics. 1st ed. Singapore: Harcouret Ara Pvt Ltd; 2000.
[3] Behman,et al. Essential paediatrics. 2nd ed. Singapore: Harcourt Brace Publishers; 1998.
[4] Catherine.E. Paediatrics. 1st ed. Philadelphia: W.B. Saunders Company; 1990.
[5] Dutta. Paediatric nursing. 2nd ed. New Delhi: Jaypee Brothers Publications; 2009.
[6] Elmen. R. Grossman. Everyday Paediatrics. 2nd ed. New York: Mc Grew Hill Company Publication; 1994.