Psychiatric Study of patients with Beta - Thalassaemia Major

Thesis

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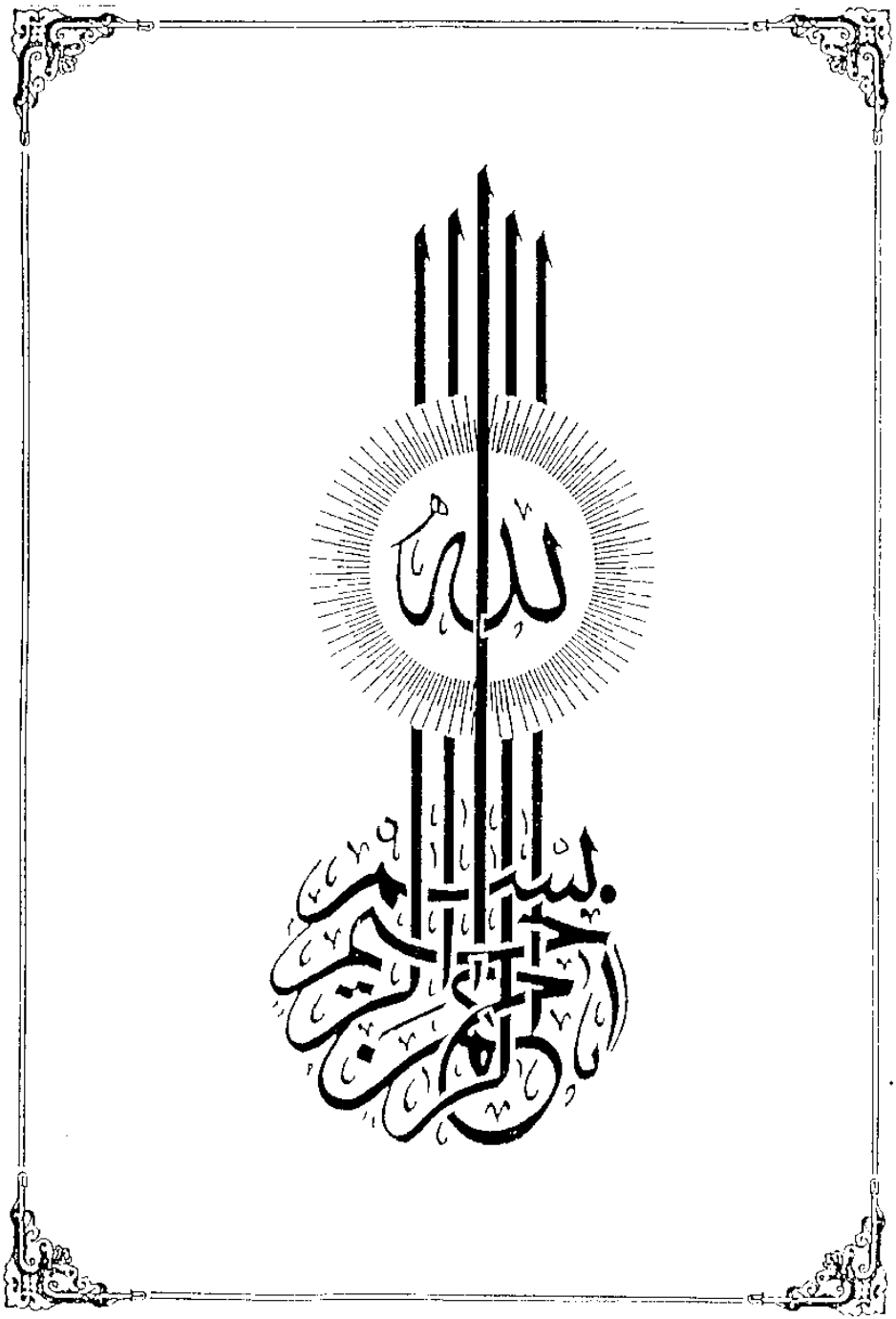
C O N T E N T S



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INTRODUCTION

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Beta-thalassaemia major is the commonest chronic haemolytic anaemia in Egypt (Sabry, 1973). Children affected by this disease are kept alive by repeated blood transfusions. Transfusion therapy may aim at keeping haemoglobin concentration at reasonable level to maintain as near as possible normal growth and development. However there are still many problems related to the clinical management of the patients due to the severity of the disease perse and the complications of blood transfusion and iron overload (Economidou, 1982).

Children with moderate to severe handicapping long term illness are at risk, not only medically, but for complex social, educational and emotional difficulties (Perrin and Gerrity 1984).

Their illness is a source of chronic stress to themselves, to their parents, and to the rest

of their families (Tsiantis et al., 1982).

The impact of illness on the family can occur in any of very many spheres: financial, somatic, behavioural, conscious and unconscious mental life, and any combination of these (Sabbeth, 1984). There is general agreement that in patients with thalassaemia, psychological problems are common (Logethetis et al., 1971).

The present work is a trial to study the effect of thalassaemia major and its prolonged therapy and follow-up on the behaviour and emotions of the affected patients and to demonstrate the psycological burden that many families experience.

REVIEW OF LITERATURE

THE THALASSAEMIAS

The Thalassaemias are heriditary haemolytic anaemias characterized by decreased or absent synthesis of one of the globin subunits of the haemoglobin molecule. Thalassaemias are named after the specific globin whose synthesis is depressed. Most cases are of the Alpha or Beta type (Kendall, 1983).

In the α-thalassaemia, decreased synthesis of the α globin results in accelerated red cell destruction because of the formation of HbH (B_4) inclusions in mature red cells (Nienhuis et al., 1984).

In B-thalassaemia, either no B chain (B^o) or small amounts of B-chains (B^+) are synthesized (Hoffbrand et al., 1980). The profound deficiency of B-chain production leads to a marked hypochromic microcytic anaemia (Ohene and Schwartz, 1980).

Excess α chains precipitate to form insoluble inclusion bodies in the marrow normoblasts (Pessas, 1963). The major factor contributing to the anaemia is haemolysis, occuring both in the bone marrow (ineffective erythropoiesis) and elsewhere, especially in the spleen. The precipitates of α chain which form, confer a rigidity to the red cells which leads to difficulty in passage through the reticuloendothelial system and to early destruction. Markedly increased erythroid activity in an unsuccessful attempt to overcome the increased rate of destruction results in enormous expansion of the bone marrow (Ohene and Schwartz, 1980).

CLINICAL FEATURES AND LABORATORY FINDINGS OF B-THALASSAEMIA

Patients with severe B-Thalassaemia are usually diagnosed between 6 months and 2 years of age. On presentation, infants usually have pallor, poor growth and development, and abdominal enlargement.

The electrophoretic patterns show 20 to 100 percent HbF, 2 - 7% HbA_2 and 0 to 80 percent HbA.

The red cells are microcytic, the Hb level may be as low as 3 - 4 gm/dl. (Nienhuis and Propper,1981).

Antenatal diagnosis:

The science of Fetology, which seeks to cure the illness in an unborn child before delivery, is considered as the frontier of pediatric medicine. A method to sample foetal blood has been detected, which can be tested for sickle cell anaemia and Beta-Thalassaemia. In the future, such technique will be routine (Kiester, 1977).

Another discovery with profound implications for foetology is that investigators were enabled to sequence human genes so that the structure and organization of some genes is now known. These observations led to studies of "natural mutations" such as the haemoglobinopathies and thalassaemias. This information in turn provided insight so that many mutations affecting the alpha - or beta - globin genes can now be detected prenatally. It seems probable that these techniques will be applicable to many other human genetic diseases. In the future, however, the greatest impact of these advances may be through their application to produce proteins, such as hormones and vaccines, which can be used to treat deficient patients (Phillips, 1983).

Management :

The management of Thalassaemia Major is straightforwards. Transfusion therapy, splenectomy when indicated and treatment with iron chelating agents to control transfusisnal iron overload (Modell and Petrou, 1983).

1. Transfusion therapy:

The mainstay of the management, remains of necessity, blood transfusions.

a) Regular blood transfusions: are needed to maintain Hb over 8 gm/dl. This usually requires 2 - 3 units every 4 - 6 weeks (Hoffbrand and Petit, 1980). Transfusions are necessary to prevent profound weakness and cardiac decompensation due to anaemia. Unless transfusions are given, life expectancy is only a few years (Nelson, 1983).

b) High transfusion program: in which the Hb is maintained at levels greater than 10 gm/dl has

many benefits. It allows children to participate fully in normal activities with their peers, decreases the degree of hepatosplenomegaly and prevents or decreases bone changes (Schwartz, 1980).

C) Recently " super transfusion": regimen has been described in which the Hb level is maintained above 12 gm/dl. The increased iron load of the augmented transfusion requirement should theoretically be partially offset by a decrease in gastrointestinal iron absorption, and freedom from anaemia should allow for completely normal grouth and development. In many patients, the supertransfusion regimen appears to be iron - sparing because little additional blood is required for " super" compared with " high" transfusion.

Apparently, normalization of Hb levels tends to decrease the erythropoietic stimulus and leads to a decrease in intramedullary volume and its associated

nutrient blood requirements (Nienhuis and Propper, 1981). Regardless of the transfusion regimen followed, washed packed red cells are preferable so as to avoid the development of antileucocytic antibodies that can give rise to febrile transfusion reactions.(Willoughby, 1977).

Prior to the initial transfusion, a complete red cell typing is done. The child has scheduled appointments for transfusions at a set interval which is individually determined. Most children require transfusions every 2 - 6 weeks. A blood sample is drawn in the morning for blood count and cross matching, an interval history and complete physical examination are done, and the transfusion is given (Schwartz, 1980).

2. Splenectomy:

Splenectomy may be of major benefit to the patient with thalassaemia major. It may be done to