STUDY OF MORBIDITY PATTERN IN CHILDREN WITH TRANSFUSION DEPENDENT THALASSEMIA

Thesis
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SUMMARY

Beta-thalassaemia represents a group of recessively inherited haemoglobin disorders characterized by deficient synthesis of the b globin chain. The homozygous state results in severe anaemia in infancy which requires regular blood transfusion.

The most important forms of thalassemia result from autosomal mutant genes that reduce the rate of synthesis of α and β chains of Hb A, designated α and β thalassemia respectively.

In Egypt, β-thalassemia is the commonest form of chronic hemolytic anemia among Egyptian children.

Thalassaemia is the most common monogenic disorder in the world. it is present in all ethnic groups. Therefore it is estimated that each year more than 5000 children are born with transfusion dependent b-thalassaemia. In the last decade the average lifespan of these patients was not more than 10years. The combination of blood transfusion and chelation therapy has dramatically prolonged the life expectancy of these patients, thus transforming thalassaemia from a rapidly fatal disease of childhood to a chronic disease compatible with a prolonged life.

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Heba Ahmed


بسم الله الرحمن الرحيم
وَأَنْزَلَ اللهُ
عَلَيْكَ الْكِتَابَ
والْحِكْمَةَ
وَعَلَّمَكَ مَا لَمْ
تَكُنْ تَعْلَمُ
وَكَانَ فَضْلُ
اللهِ عَلَيْكَ
عَظِيمًا
صدق الله العظيم
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List of Abbreviations

γ	*Gamma*
α	*Alpha*
β	*Beta*
ζ	*Zeta*
ε	*Epsilon*
Hb	*Hemoglobin*
IV	*Intravenous*
SC	*Subcutaneous*
TLC	*Total leucocytic count*
Plt	*Platelet*
T. bil	*Total bilirubin*
D. bil	*Direct bilirubin*
EF	*Ejection fraction*
SD	*Standard deviation*
LVEF	*Left ventricular ejection fraction*
DFO	*Defereprone*
RDW	*Red cell distribution width*
rHuEPO	*Recombinant human erythropoietin*
CPT	*Carnitinepalmitoyltransferase*
OCTF	*Organic carnitine transporter family*
IGF-1	*Insulin growth factor 1*

INTRODUCTION

Thalassemia is a group of chronic hereditary hemolytic anemias particularly common in persons of Mediterranean, African, and South Asian ancestry in which there is decreased synthesis of one or more hemoglobin polypeptide chains ***(Palma et al., 2005).***

Generally, the estimated prevalence is 16% in people from Cyprus, 3-14% in Thailand and 3-8% in populations from Bangladesh, China, India, Malaysia and Pakistan. A very low prevalences has been reported from people in Africa (0.9%) with those in North Africa having the highest prevalence and northern Europe 0.4% ***(Wambuas et al., 2006).***

Depending on the genes involved, thalassemia is classified as Alpha-Thalassemia or B Thalassemia which is further classified into B thalassemia major and B Thalassemia intermedia. Thalassemia major presents typically within the first year of life and patients subsequently require ongoing transfusions to survive, Thalassemia intermedia presents late in life and patients may be transfusion independent or require only sporadic transfusion ***(Forget et al., 2005).***

Quality and duration of life of transfusion-dependent B Thalassemia patients have been transformed over the last ten years with life expectancy increasing well into the third and the forth decades. Nevertheless, the life prolongation

discloses several complications partly due to the underling disorders, partly related to the conventional treatment with blood transfusions and to iron overload ***(Borgna et al., 2003).***

Complications of thalassemia major are mostly due to iron overload caused by frequent blood transfusions, and include heart failure, infections, hypogonadism , infertility, diabetes mellitus and hypothyrodism ***(Olivieri, 1999)***.

Cardiac complications remain the most important in determining the survival of B -Thalassemia major patients, it should be emphasized that they are not restricted to the effect of anemia and iron loading although they are by far the most important factors ***(Piga and Longo, 1997).***

Thalassemia patients have an increased risk of infections because of splenectomy, iron load and blood born infections particularly viral (**Adam ,** ***1998).***

Endocrine abnormalities are among the common complications of thalassemia despite good chelation therapy. Delayed puberty and defective function of the hypothalamic /pituitary axis occurs in approximately 56% of both male and female patients ***(Cohen, 2000).***

Osteoporosis is a multi- factorial complication and is very common in adult thalassemia patients of both sexes,it require an adequate management in order to prevent progressive disease and fractures ***(Locatelle, 2004).***

AIM OF THE WORK

The aim of this work is to study the morbidity patterns of transfusion-dependent thalassemia patients, and compare the outcomes of these patients in relation to age of onset, type, duration and compliance to iron chelation therapy and frequency of blood transfusion.