Population Screening Programme of Thalassaemic Patients in Barpeta Region of Assam

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***Abstract*—The β-thalassemias are characterized by a very heterogeneous group of inherited mutations causing abnormal expression of globin genes, leading to total absence or quantitative reduction of synthesis of β-globin chains (1–3). This disease is frequent in the Mediterranean area, Middle East, Africa and Asia. More than 200 different mutations have been identified in β-thalassemia patients, including deletions of the β-gene region, stop codons leading to premature termination of a non-functional β-globin chain, mutations suppressing correct maturation of the β-globin RNA precursor, most of all need regular blood transfusions.**

In this present study we performed a population screening programme of thalassaemic patients in Barpeta region of Assam. In this programme we found 82 E-Bata, 18 Beta among 100 patients.

***Index Terms*—β-Thalassaemia, β-globin chain, Blood transfusion.**

I. INTRODUCTION

The β-thalassemias are characterized by a very heterogeneous group of inherited mutations causing abnormal expression of globin genes, leading to total absence or quantitative reduction of synthesis of β-globin chains **(1–3).** This disease is frequent in the Mediterranean area, Middle East, Africa and Asia. More than 200 different mutations have been identified in β-thalassemia patients, including deletions of the β-gene region, stop codons leading to premature termination of a non-functional β-globin chain, mutations suppressing correct maturation of the β-globin RNA precursor, most of all need regular blood transfusions. **(1–3,4,5).**

It is firmly established that increasing the production of γ-globin leads to a decrease in the imbalance between β and non-β-chains and the consequent reduction of haemolysis. **(6–9),** including the very interesting finding that a group of genetic mutations, known as hereditary persistence of fetal hemoglobin (HPFH), are associated with high levels of HbF in adults **(10–13)** resulting in a mild phenotype. Most of the HPFH patients homozygous for β-thalassemia do not need blood transfusion. Therefore, there has been considerable interest in recent years to solve this problem in finding ways of increasing production of HbF and reactivation of the γ-globin genes (14–23).

II. MATERIALS AND METHODS

A. Study groups:

Patients with HPLC-screened documented Beta thalassaemia, HbE-beta thalassaemia genotypes have been considered in this screening programme. Total 100 patients were evaluated in this screening programme.

III. RESULT

In our population screening study, we evaluated total 100 patients. Among which 82 patients with Hb-E-beta and 18 patients with Beta thalassaemia were observed. This was clearly depicted in Chart 1. We also evaluated the male and female percentage among Beta and Hb-E-beta Thalassaemic patients. Which were clearly depicted in Chart 2 and Chart 3

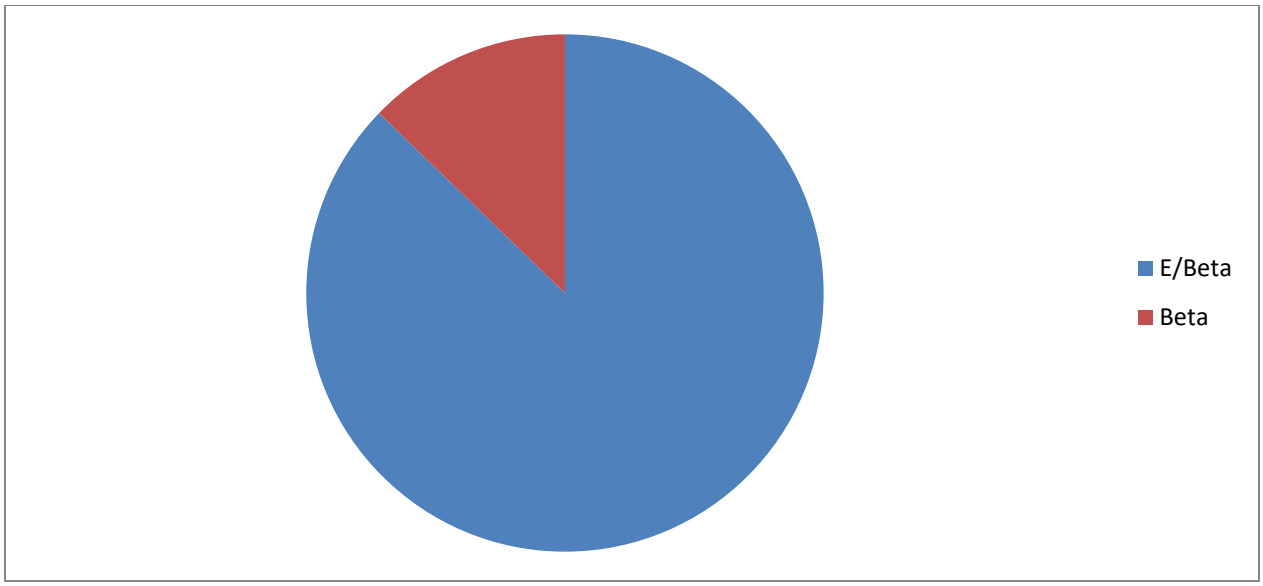

Chart 1: Population Screening of Beta, E-Beta, among total 100 thalassaemic patients in Barpeta region of Assasm.

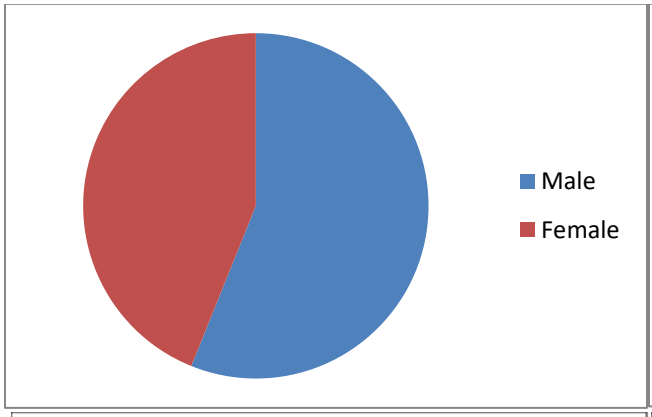

Chart 2 Male and female percentage among 82 E-Beta Thalassaemic Patients.

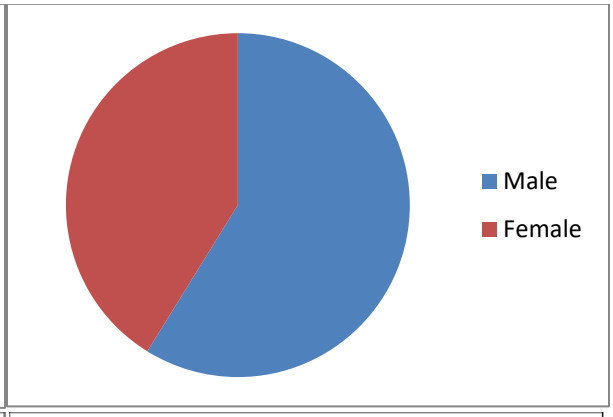

Chart 3 Male and female percentage among 18 Beta Thalassaemic Patients.

IV. DISCUSSION

β-thalassemias (β-thal) are common inherited red cell disorders characterized by absent or reduced synthesis of β-globin chains. Despite extensive knowledge of the molecular defects causing β-thalassemia, less is known about the mechanisms responsible for the associated ineffective erythropoiesis and reduced red cell survival (24-31). Increased levels of reactive oxygen species (ROS) have been reported to contribute to the anemia of β-thalassemia, although the effects of ROS have not been fully defined (24, 26-31). Exogenous anti-oxidant molecules might represent complementary therapeutic strategies to counteract the toxic effects of ROS in β-thalassemia. However, few of them have been shown to beneficially affect *in vivo* β-thalassemic red cell features and/or thalassemic ineffective erythropoiesis *in vivo*. (27-31).

In our present population screening study, we observed that the occurrence of E-beta thalassaemia was very higher than Beta thalassaemia in Barpeta region of Assam which was clearly mentioned in chart 1 and other two charts, Chart 2 and 3 we observed that both E-Beta and Beta cases the effected males were always higher then effected females.

V. CONCLUSION

Beta Thalassemia is an inherited disorder in which either very few or no red blood cells are produced by the bone marrow after infancy. The treatment is monthly whole blood transfusions and the use of a drug which is extremely toxic and cannot be used with children. The disease dramatically impacts the sufferers' quality of life and often results in death around the age of puberty. Because it is more common in less developed countries where it is virtually impossible for anyone other than the very wealthy to obtain regular supplies of clean whole blood for the required transfusions, the fatality rate is high. Even if the patient is able to obtain monthly transfusions and is able to afford the drugs to treat the disease, he or she is constantly anemic and lacking of energy.

In our present study we conclude that in Assam Barpeta region the occurrence of E-Bata thalassaemia was higher than Beta Thalassaemia and the percentage of effected male patients were higher than the effected female patients.

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B. Competing Interests Statement:

The authors declare that they have no competing interests.

C. Data Sharing Statement:

We cannot share any unpublished data with other laboratory or person.

D. Patients Consent Statement:

The signed consent from all the patients were taken before test was performed and kept them as official documents. In case of any unusual condition it will be presented in front of the concerned person.

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