PREFACE

Hemoglobin is a molecule present in red blood cells responsible for Oxygen transportation. Adult hemoglobin is a hetero-tetramer composed of two $\alpha$- and two $\beta$-globin chains ($\alpha_2\beta_2$), each of which contains a heme molecule capable of binding Oxygen and facilitating Oxygen transport. Thalassemias are the conditions of abnormal hemoglobin production due to deletions or mutations on globin genes. The two most clinically relevant thalassemia syndromes are the $\alpha$- and $\beta$-thalassemias. Alpha thalassemia is caused by deletion or mutation affecting either one or more of the duplicated $\alpha$-globin genes, located on chromosome 16, whereas $\beta$-thalassemia is caused by point mutations in $\beta$-globin gene, located on chromosome 11p15.5, leading to an absence or decrease in globin chain production from the affected genes.

The thalassemias are inherited in a Mendelian recessive fashion. The severe, homozygous form of the disease is called thalassemia major and the carrier state (in which only one defective globin gene is inherited) is called the trait. The disease is very heterogeneous from the clinical point of view; many patients are encountered who fall between these extremes. These latter disorders are called ‘thalassemia intermedia’.

Majority of people with $\beta$-thalassemia trait is asymptomatic, they may not be aware of their carrier state. It may be associated with mild or no anemia but with reduced mean corpuscular volume (MCV), mean corpuscular hemoglobin (MCH) values and an elevated hemoglobin $A_2$ ($HbA_2$) level. On the other hand, $\beta$-thalassemia major patient are severely anemic and need lifelong blood transfusion for their survival.

The prevention of homozygous condition can be achieved through the detection, counseling and education of heterozygous carriers. One way of achieving this goal is to screen the population at risk. The screening of carriers can be done by osmotic fragility test (NESTROFT), red cell morphology, red cell indices and different discriminant formulae, estimation of $HbA_2$ by cellulose acetate electrophoresis, or high-performance liquid chromatography (HPLC) and DNA analysis.

The present study aimed to screen out extended families of $\beta$-thalassemia major patients who reported at Govt. Civil Hospital, Dhule for routine blood transfusion. The long-term objective of this research work is to educate the affected
families about all concerned information of thalassemias and try to reduce the incidence of this dreaded disease in the affected families. This type of study will definitely help not only the affected families but also to the nation for making a national control design to reduce the incidence of disease at some extent.