# SUMMARY

β-Thalassemia is considered the most common genetic disorder worldwide, it occurs in a particulary high frequency in abroad belt extending from the Mediterranean basin throught the Middle East, and abundance in Egypt.

The thalassemias are a group of genetic (inherited) blood disorders that share in common one feature, the defective production of hemoglobin. There are many different disorders with defective hemoglobin synthesis and, hence, many types of thalassemia. About 3% of the world's population (180 million people) carry β-thalassemia genes.

The present study was carried out in the Biological Application Department of Nuclear Research Center, Atomic Energy Authority and Microbiology Department and Hematology unit of Pediatrics Department, Faculty of Medicine, Zagazig University. This study included 75 patients, their age ranged from 3-15 years and divided into two groups:

**First group:** Consists of 50 children (33 males & 17 females) had β-thalassemia, positive consanguinity present in 27 patients while positive family history present in 17 patients. All of patients were blood transfusion dependant.

**Second group:** Consists of 25 healthy children (17 males & 8 females) as a control group.

The aim of this study to determine the biochemical changes in serum and study some of mutations in Intron-1 of β-globin gene of β-thalassemic children in our locality Sharkia province.

The hematological parameter such as Hb concentration, serum iron, TIBC, ferritin and Hb electrophoresis were done to both control and patient groups. As well as the molecular studies, extraction of DNA, amplification by using PCR and digestion by two restriction enzymes NLaIII and BsmA1 were carried out to two groups.

In this study the concentration of Hb conc. and TIBC were decreased, which considered a more highly significant decreased. On the other hand, the levels of serum iron, serum ferritin were increased, this increased were more highly significant.

The hemoglobin electrophoresis was carried out for 41 patients who had stayed without blood transfusion for two months, there were a pronounced increased in the values of HbF and HbA2 this increased being more highly significant. On the other hand, the level of HbA was decreased and being more highly significant decreased.