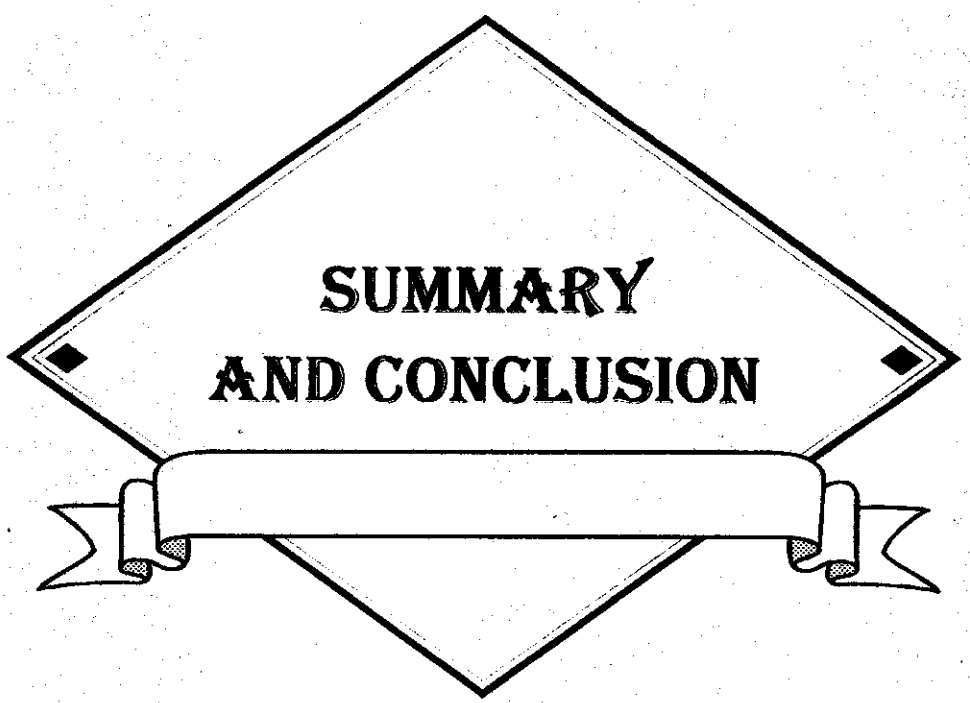
SUMMARY
AND CONCLUSION

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Thalassemia genes are remarkably widespread. They are the most prevalent of all human genetic diseases. The frequency of β-thalassemia trait, which is the commonest hemolytic anemia, in Egypt is estimated to be as high as 13% of the whole population. It represents a major health problem in our country which has to be managed on a national scale. Premarital and prenatal diagnosis of carriers and cases had succeeded remarkably in lowering the incidence of new cases in Cyprus. Also a well organized national program in this island had improved much the quality of life with prolongation of the life expectancy of these patients who can live almost normally.

Supertransfusion and intensive chelation therapy with desferal is still the corner stone for management of these patients, but the poor compliance and high cost are major problems. Other oral iron chelators are tried with much controvertial results. This therapy is considered a symptomatic one. The real curative treatment available is bone marrow transplantation with all its hazards and obligations. Recently intrauterine transplantation with stem cells from healthy compatible cord blood is tried with encouraging results. Fetal hemoglobin induction is also tried. Splenectomy seems to be mandatory at about 6 years of age. Other supportive and supplementary therapeutic measures are to be considered e.g. vitamins particularly vit. C and folic acid, minerals particularly Ca, Zn and Mg., hormonal replacement and vaccination particularly hepatitis B and penumococcal vaccines.

A major problem added to the every day encountered thalassemia is the disturbed trace element metabolism. The exaggerated red cell destruction and bone marrow hyperactivity will disturb element metabolism. The life-long use of the iron chelators especially desferal raises the need for studying their possible effect on these elements. This drug has a specific affinity for trivalent cations Fe^{+++} and Al^{+++}, hence its use for their chelation. Its effect on other divalent cations is still a subject of much controverse.

The aim of this work is to determine the effect of the most commonly used and effective iron chelating agent (desferrioxamine) on Fe, Al, Pb, Zn, Cu, Ca, Mg and Mn.

Subjects of this work were devided into 3 groups :

1- Seventy five thalassemic children receiving irregular therapy.

2- Twenty five thalassemics receiving regular therapy with the pump.

3- Twenty five normal healthy children as a control group.

A detailed history and thorough physical examination was carried out for each child. Blood samples were withdrawn from each one just before blood transfusion for the following investigations - complete blood picture, hemoglobin electrophoresis, and serum level of Fe, Al, Pb, Ca, Mg, Zn, Cu and Mn. Statistical analysis of data shows that the drug has a significant chelating effect on both Fe and Al. Regularity in administration, with the pump, is essential for Fe but not Al chelation. That is why it is an effective therapy for Al & Fe toxicity.

Both the drug and the disease seem to show no significant effect on serum lead level. Their possible lowering effect on Ca is modified by the

routine use of Ca supplementation in the therapeutic regimen of our patients. Thalassemia is reported to lower serum Zn and elevate Cu. This effect is modified in our study by the repeated blood transfusion which is rich in Zn. Both the drug and the disease was reported to lower serum Mg level which is not the case in our study. Also the drug showed no significant effect on serum Mn level.

Hb % in all diseased patients are low indicating poor compliance with therapy. HbA2 is within normal range possibly due to blood transfusion or the genetic type in our country.

Correlation studies show the -ve relation between Ca and Mg ions in the irregular group which are believed to be antagonists. Splenectomy seems to raise Hb% in irregular group with a rise in serum Ca level.

Unfortunately there is little updated work done in this essential aspect of management of this disease. This raises the importance of further evaluation and repeated follow-up of our thalassemics.

Conclusion :

Thalassemia is the commonest genetic disorder and the commonest hemolytic anemia in Egypt. It is widely spread in our country with a carrier rate up to 13% of the general population. It is to be considered in every case of microcytic anemia that resists iron therapy. Antenatal diagnosis and screening with an integrated preventing programs are essential for early detection and prevention of this disabling disease. Supertransfusion, intensive chelation therapy with all its difficulties is still the corner stone for management. Desferal is the main internationally accepted iron chelator

although others are tried orally. Bone marrow transplantation is the only available curative therapy. Gene therapy is the hope for real etiological, rather than symptomatic, treatment.

Minerals are essential partners in many metabolic and enzymatic activities of the human body and their importance was discovered as early as the ancient civilizations. Disturbed mineral metabolism and balance is an important sequel of thalassemia major with its life long therapy. This study is carried out to throw lights on this important aspect of the disease. The essential cations Fe, Al, Pb, Ca, Mg, Zn, Cu and Mn were studied. Desferal succeeded to lower serum level of Fe and Al but its effect on other minerals is not manifest. Supplementation with Ca has successfully corrected the lowering disease effect. Its effect on serum Zn is corrected by blood transfusion which is a rich source of Zn. Hb% in our patients is still under corrected which raises the importance of a more integrated wide spread team work efforts allover the country for better control of the disease.