SUMMARY

Glycogen storage disease (GSD) is caused by inherited defects in one of the enzymes involved in the synthesis or degradation of glycogen. Liver and muscle, with abundant glycogen, are the most commonly and seriously affected.

The overall GSD incidence is estimated 1 case per 20 000 - 43 000 live births.

There are over 12 types and they are classified based on the enzyme deficiency and the affected tissue.

Hypoglycemia and hepatomegaly is the primary manifestation of the hepatic glycogenoses, whereas weakness and muscle cramps are the predominant features of the muscle glycogenoses.

Growth retardation is a well-known problem for patients suffering from glycogen storage diseases (GSDs). However, the underlying mechanism, and therefore the management of growth impairment in these patients, remains controversial. Hyperlacticacidemia, recurrent hypoglycemia, growth hormone (GH) and/or insulin-like growth factor (IGF) deficiency, GH and/or IGF resistance, decreased insulin and increased cortisol secretions have all been suggested to explain growth retardation in GSDs.

This search aimed at study endocrine and metabolic variables that affect the growth in patients with glycogen storage diseases receiving standard dietary therapy.

This is done by Observational study for twenty-five patients with GSDs, age range 1-12 years, were investigated on their usual dietary regimens. Data on height, weight, BMI and other laboratory investigations as CBC, liver enzyme, lipid profile and study of GH, IGF1 concentration profiles and compared to twenty healthy subjects then the results statistically analyzed.

The results show that these patients although they had therapeutic dietary regimen they still shorter than average and also they had increased BMI and

abdominal circumference. They show hepatomegaly in most cases and splenomegaly in few cases on our clinical examination .Also we found the liver enzymes of cases and their lipid profile were elevated. The endocrinal profile of our cases demonstrated GH insensitivity in form of increased GH and decreased IGF1in general in our cases. The shorter the child and the more increased in BMI, the more increased in the level of GH and the more decreased in the level of IGF1.

In the present study, this pattern in the growth of those affected children who received dietary therapy since the birth is explained with the exposure of their cells for a state of chronic starvation and metabolic acidosis which present as a result to the failure of the liver to release glucose from glycogen and the persistent hypoglycaemic stimulus that produces lactic acidosis through the intact pyruvate pathway. These two conditions were found to cause disturbance in the GH-IGF1axis in form of GH insensitivity.

Therefore, this study demonstrates that these patients are suffering from failure to reach to optimal response of treatment and the cause needs to be understood through further investigations.

Also the data from this study will not support the use of GH, that GH has been occasionally used therapeutically in GSDs in an attempt to improve poor growth, but with little success, as a therapeutic strategy.