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MEDICAL-NUTRITIONAL INTERVENTION IN A JORDANIAN CHILD WITH GLYCOGEN STORAGE DISEASE TYPE IIIA: CASE REPORT

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Abstract

Background: *Glycogen storage disease (GSD) type IIIa is a rare inborn error of metabolism characterized by a deficiency in glycogen disbranching enzymes. Nutritional intervention is a cornerstone in the medical care plane.* ***Case presentation:*** *A 2-year-old Jordanian male, who is known to have GSD IIIa since he was 4 months was admitted because of infection. The child was on a special diet (small, frequent meals of complex carbohydrates and protein, avoiding simple sugars and fasting is prohibited). The child showed good activity level and a good appetite.* ***Method:*** *The medical-nutritional intervention of GSD IIIa was evaluated by retrograde reviewing the child BMI, blood and biochemical tests on presentation and a month later visit.* ***Results:*** *The biochemical tests included: blood glucose, urea, creatinine, cholesterol, triglycerides, albumin, total bilirubin, aspartate amino transferase (AST), alanine aminotransferase (ALT) and WBCs were decreased after nutritional intervention, however, the RBCs blood test was increased. On presentation, the child's weight and height were documented as above the 15th and at 97th percentile respectively for his age, no change after the one month later visit was observed.* ***Conclusion****: The biochemical and blood tests improved at the one-month follow-up visit vs. baseline. The individualized medical-nutritional intervention is a cornerstone in the management of GSD IIIa as part of a comprehensive medical care process.*

key words: *Glycogen storage disease IIIa, inborn error of metabolism, medical-nutritional intervention.*

Background and Aims

Glycogen storage disease type III (GSD III) also known as Cori disease, is a part of a rare group of inherited enzyme defects that affect the glycogen metabolism to produce glucose. It is an inborn error of metabolism caused by autosomal recessive deficiency in glycogen disbranching enzymes [1]. It is a disease of variable clinical severity affecting primarily the liver and muscle tissue. Internationally, the frequency and incidence GSD III is 1 children per 100,000 live births [2-4]. Deficiency of the enzyme in liver and muscle tissue produces a variant known as GSD IIIa which is more common, whereas the GSD IIIb is another variant caused by a deficiency of the enzyme in the liver only without muscle involvement [1,5].

The GSD III shows diverse clinical presentations. In infancy and early childhood, it

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presents with recurrent infection, hypoglycemia, hepatomegaly and growth retardation is not unexpected [5]. On the other hand, during adulthood, it presents only with asymptomatic hepatomegaly, myopathy or occult liver disease [6]. In a small portion of patients with GSD III end stage liver disease or cirrhosis may occur due to chronic fibrosis [7-9]. Generally, fasting hypoglycemia improves with age and hepatomegaly frequently regresses [10-11].

Case presentation

In this case study, acute management of the child is beyond the scope of this case however, medical-nutritional intervention of GSD IIIa was evaluated by retrograde reviewing the BMI, blood and biochemical tests on presentation and a month later visit for a 2-year-old Jordanian male who is known to have a GSD IIIa since he was 4 months. His parents are relatives with no known family history of GSD IIIa. He had been breastfed for 4 months. The child is on a special diet (small, frequent meals of complex carbohydrates and protein, avoiding simple sugars and fasting is prohibited). The child showed good activity level and a good appetite. The child's parents are eager to compresence and learn more about their child's case from nutritional point of view so they referred to department of clinical nutrition to know more about their child rare disease.

The weight and height were reported by the child's parents (11.3 Kg and 98 cm) respectively. A series of biochemical tests that conducted during hospitalization including Urea, Creatinine, Cholesterol, Triglycerides, Albumin, total Bilirubin, Aspartate Aminotransferase (AST) and Alanine Aminotransferase (ALT). Moreover, the blood test included Hemoglobin, WBCs and RBCs. A written informed consent was obtained from child parents after the nature of the case study had been explained.

Table 1. The blood test on presentation and one month later after nutritional intervention.

Blood test	On presentation	One month later	Normal range
Hemoglobin	13.9 g/dl	13.8 g/dl	10-14 g/dl
WBCs	24.7 x 10^3 /µl	10.2 x 10^3 /µl	(3.8-10.8) ×10^3 /µl
RBCs	3.79 x 10^6/µl	4.66 x 10^6/µl	(4.3-5.8) ×10^6/µl

After retrograde reviewing of child data on presentation and on a month later visit, the child's weight and height were above the 15th and at 97th percentile for his age. When reviewing them, they were not changed after one month from presentation. This indicates normal growth with no evidence of stunting or growth retardations according to the WHO growth charts [12]. However, infants with GSD IIIa usually have high risk of growth retardation [13] making the long-term monitoring of growth is important by using standard growth charts for age in order to detect any changes in growth parameters and evaluate the nutritional related qualitative and quantitative risk by referring to medical specialist. The blood tests on presentation and at a month later visit after nutritional intervention are shown in Table 1. The WBCs test (24.7 x 10^3 /µl vs. 10.2 x 10^3 /µl) was lower after nutritional intervention, on other hand, the RBCs test (3.79 x 10^6/µl vs. 4.66 x 10^6/µl) was increased after intervention.

The blood test showed normal level of hemoglobin indicating a good nutritional status as no risk of anemia. Whereas, high white blood cell count (leukocytosis) related to active infection on presentation [14] which was associated with hypoglycemia (62 mg/dl) that resolved after infection is resolved through medical intervention. A close blood glucose monitoring is required during course of illness and the routine immunization should be

warranted to know if any is missing and decrease risk of infection [11].

The biochemical tests on presentation and on a month later visit after nutritional intervention are given in Table 2. The following biochemical tests were different after nutritional intervention: Random blood glucose (62 mg/dl vs. 77 mg/dl), Urea (50 mg/dl vs. 19 mg/dl), Creatinine (0.9 mg/dl vs. 0.5 mg/dl), Cholesterol (260 mg/dl vs. 143 mg/dl), Triglycerides (243mg/dl vs. 171mg/dl), Albumin (3.7 g/dl vs. 3.3 g/dl) and Bilirubin total (0.67 mg/dl vs. 0.5 mg/dl). The Aspartate aminotransferase (AST) and Alanine aminotransferase (ALT) were also lower after nutritional intervention (790 U/L vs. 226 U/L) and (239 U/L vs. 133 U/L) respectively. As shown above, the child had hypercholesterolemia and hypertriglyceridemia on presentation.

Table 2. The biochemical test on presentation and one month later after nutritional intervention.

Biochemical test	On presentation	One month later	Normal range
Random blood glucose	62 mg/dl	77 mg/dl	70-140
Urea	50 mg/dl	19 mg/dl	15-45 mg/dl
Creatinine	0.9 mg/dl	0.5 mg/dl	0.5- 1.4 mg/dl
Cholesterol	260 mg/dl	143 mg/dl	< 200 mg/dl
Triglyceride	243 mg/dl	171 mg/dl	30-200 mg/dl
Albumin	3.7 g/dl	3.3 g/dl	3.4- 5 g/dl
Bilirubin, total	0.67 mg/dl	0.5 mg/dl	< 1.5 mg/dl
Aspartate aminotransferase (AST)	790 U/L	226 U/L	0 – 60 U/L
Alanine aminotransferase (ALT)	339 U/L	133 U/L	0-50 U/l

This is consistent with other studies results, where 40% of those with GSD III have hyperlipidemia [15,16]. However, not all studies confirm this finding [17]. The hypertriglyceridemia and hypercholesterolemia were correlated negatively with age, and may reflect increased severity of hypoglycemia in this younger population [18-20]. The child urea and creatinine level were typically normal on presentation. Whereas, elevated hepatic transaminase concentrations with more than 500 U/L aspartate aminotransferase (AST) concentrations were also observe [3,20]. Although Serum level of AST and ALT are markedly elevated in the first decade of life, it tends to decrease significantly thereafter [5]. In general, Hepatomegaly among individuals with GSD III may improve with age and usually resolve after puberty [3]. All previous biochemical tests were improved after one month of medical-nutritional therapy which aimed to decrease the workload of the liver by providing a diet with lower simple sugar, high in complex carbohydrate and protein and normal fat with bedtime snack and cornstarch to enhance gluconeogenesis and prevent hypoglycemia. Follow up every 6 months of serum AST, ALT, bilirubin and albumin to yearly detect the extent of hepatic damage monitor progression of liver failure and routine immunizations should be offered including hepatitis B [3]. The plan of medical-nutritional intervention of GSD IIIa in general should be individualized, no specific therapy exists. However, it is mainly based on symptomatic and nutritional therapies.

Conclusion

These results emphasize the importance of the collaboration among multidisciplinary

medical care professionals including clinical nutritionist; to establish an individualized medical-nutritional intervention in children of GSD IIIa to ensure the patients best care and outcome.

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