ECRONICON OPEN ACCESS

EC PAEDIATRICS
Case Report

Mauriac Syndrome, Rare Complication of Uncontrolled DM Type I, Case Report

Iman A Al Mukhtar[1]*, Nesreen AlZaki[2], Fadel AlBasarah[3], Zainab AlAbbas[3], Fatimah AlAbbad[3] and Ali AlDhamen[3]

[1]*Pediatric Critical Care Consultant, Qatif Central Hospital, Qatif, Saudi Arabia*

[2]*Pediatric Endocrinology Consultant, Qatif Central Hospital, Qatif, Saudi Arabia*

[3]*Pediatric Resident, Qatif Central Hospital, Qatif, Saudi Arabia*

***Corresponding Author:** Iman A Al Mukhtar, Pediatric Critical Care Consultant, Pediatric Department in Qatif Central Hospital, First Health Cluster Eastern Province, Ministry of Health Saudi Arabia, Saudi Arabia.

Received: September 30, 2020; **Published:** October 12, 2020

Abstract

Mauriac syndrome is a rare complication of uncontrolled diabetes mellitus with growth failure, and hepatomegaly with or without association of retinopathy and nephropathy.

We hereby report a case of twelve-year old boy, known to have DM type I since age of six years. He has been poorly compliant to his medications as well as his follow up appointments, with frequent admissions to intensive care unit due to severe diabetic keto-acidosis, DKA.

As a result of his poor disease control, due to complicated socioeconomic status, he developed the following complications of growth failure, hepatomegaly, nephropathy and intellectual disability, which is consistent with typical manifestation of Mauriac syndrome.

Mauriac syndrome should be considered in any patient with poor glycemic control and aim to avoid it by achieving ideal glycemic target.

Keywords: *Mauriac Syndrome; Diabetes Mellitus Type I; Diabetic Ketoacidosis DKA*

Introduction

Mauriac syndrome is one of the rare complications of type I diabetes mellitus.

The condition was descried by Mauriac in 1930 as "hepatic glycogenosis". It is a condition of hepatomegaly, growth attenuation, cushingoid features and abnormal glycogen store. This condition occurs more in poorly controlled patients [1].

The pathophysiological background behind this condition is not well understood. Growth attenuation thought to be linked to a defect in response of hepatocyte receptors to growth hormone secondary to low circulating insulin. This will decrease IGF-1 level and then affect the growth [2].

Hepatomegaly has been linked to excessive glycogen deposition in hepatocyte. Genetic cause of Mauriac syndrome has been reported. It is linked to a mutation in PHKG2, that when combined with extreme hyperglycemia will inhibit glycogenolysis, and then lead to hepatomegaly [3].

Despite the advancement of long acting insulin therapy, Mauriac syndrome is still reported. In this case, we are reporting an adolescent patient diagnosed with diabetes mellitus type 1 which was poorly controlled with multiple admission due to DKA. The social background of him play a major role in the development of DM type 1 complications including Mauriac syndrome.

Case Presentation

This is a 12-year old Saudi male, diagnosed with DM type I at the age of six years. He presented with abdominal pain, vomiting and difficulty of breathing due to metabolic acidosis and hyperglycemia associated with a prolonged history of polyuria, polydipsia for around a one-month duration prior to presentation. His initial HbA1C was 20%.

He was diagnosed as DM type I and started on SQ insulin injections then was scheduled for regular follow up appointments at the endocrinology clinic, nutrition specialist, diabetic educator clinic and social support clinic.

His initial growth parameters were as follows:

- Weight: 20 kg (25th percentile)
- Height: 120 cm (25th percentile)
- BMI: 13.8.

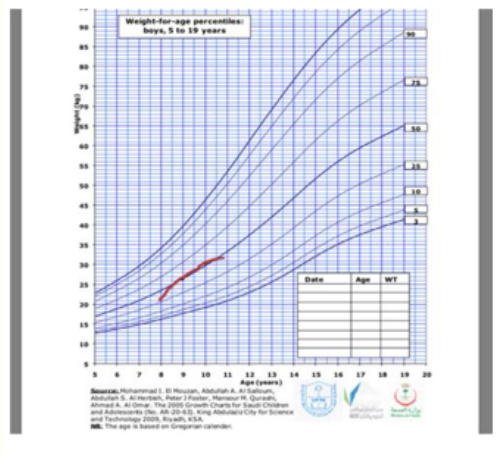

Figure 1

Subsequently, patient had frequent admissions to the intensive care unit with diabetic ketoacidosis due to poor compliance to medications, diet, follow ups. He had an average of three to five admissions per year to PICU presenting with severe DKA in addition to frequent admissions and ER visits with hyperglycemia.

His subsequent growth parameters measurements were tracked and showed slow progression of his height 2 - 3 cm/year dropping in centiles (currently below 3rd centile) and weight on the same percentile (25th)

Latest measurements were

- Weight: 27 kg.
- Height: 130 cm.

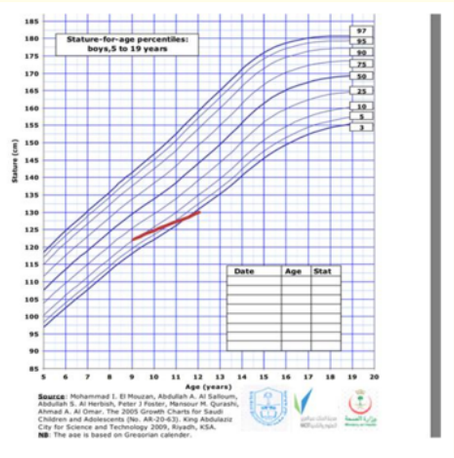

Figure 2

Physical examination reviled: undernourished boy, looks short, has protruded abdomen with fatty wastes, had hepatomegaly, liver was palpable 4 cm below costal margin, total span was 16 cm.

Tanner stage I, SMR prepubertal.

He was showing signs of growth retardation, failure to thrive and cushioned features.

Over a six years period, his overall condition was showing multisystemic involvement including a deterioration in cognition and school performance, development of hepatomegaly, worsening kidney function in addition to worsening vision as well.

Ophthalmology evaluation showed bilateral cataract and still on follow-up for possible surgery.

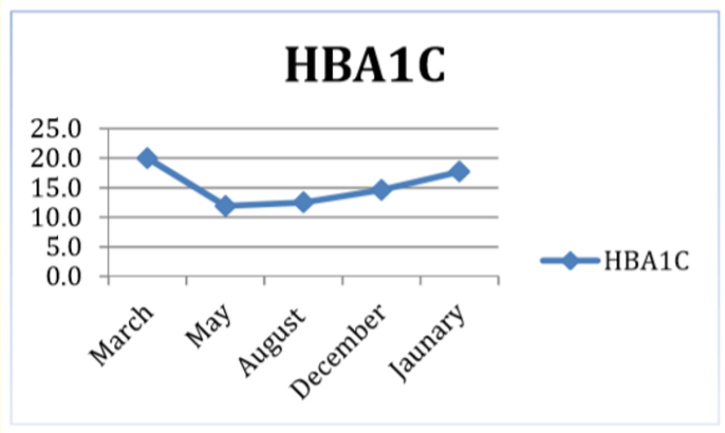

Figure 3: *HBA1C trends: Indicating poor sugar control and poor compliance to insulin injections.*

Laboratory	
Complete Blood Count CBC	
WBC	18.6
RBC	4.68
HGB	12.9
WBC	76
MCV	27
MCH	12
RDW	268
PLT	18.6
Renal function test and electrolytes	
Na	135
K	4.7
CL	91
Urea	9.8
Cr	82
Albumin	4.4
Ca	8.8
PO_4	5.2
Liver function test	
ALT	113
AST	22.7
D.Bili	1.9
T.Bili	3.6
GGT	136
Protein	87
Lipid profile	
Cholesterol	347
HDL	23
LDL	118
Triglycine	1175

Table 1

Abdominal Ultrasonography: showed mild hepatomegaly with grade 2 hepatic steatosis (Diffuse fatty liver infiltration) with renal findings consistent with diabetes-related renal papillary necrosis.

Patient was already seen by a multidisciplinary team including pediatric endocrinology, nephrology, ophthalmology, child protective services and child abuse as well as neglect committees and social workers to take over his socioeconomic and financial support, trying to

improve his quality of life and to reattach him to the health care and follow ups he needed.

Eventually he had all the clinical manifestations of Mauriac Syndrome attributable to poor glycemic control.

Reversal of these manifestations was not achieved.

Discussion

Diabetes mellitus describes a complex of metabolic disorder characterized by chronic hyperglycemia resulting from defects in insulin secretion, insulin action or both. A marked elevation of the BGL confirms the diagnosis of diabetes, including a random plasma glucose concentration ≥ 11.1 mmol/L (200 mg/dL) or fasting plasma glucose ≥ 7.0 mmol/L (≥ 126 mg/dL) in the presence of overt symptoms or hbA1c >= 6.5% [4].

ISPAD recommend regular self-monitoring of glucose (using accurate fingerstick blood glucose measurements, with or without continuous glucose monitoring [CGM] or intermittently scanned CGM [is CGM]), in addition to intensive insulin regimens delivered by combinations of multiple daily injections or pump therapy with differential substitution of basal and prandial insulin to achieve optimal metabolic control of targeted HbA1C of < 7% [5].

Improvements in glycemic control by intensive insulin treatment as well as healthy eating principles reduce the risks of acute and long-term complications. The optimal macronutrient distribution varies depending on an individualized assessment of the young person. As a guide, carbohydrate should approximate 45% to 50% of energy, Fat < 35% of energy (saturated fat < 10%) and Protein 15% to 20% of energy [5].

Mauriac syndrome was first described by Mauriac in 1930 in children with T1DM. It is characterized by hepatomegaly, growth failure, delayed puberty, and cushingoid features [6]. Other clinical features include protuberant abdomen, proximal muscle wasting elevated liver enzymes and dyslipidemia. However, the single presenting feature of Mauriac syndrome can be glycogenic hepatopathy in both adults and children. Glycogenic hepatopathy is an underrecognized and uncommon complication of poorly controlled DM type 1 manifested by hepatomegaly, abdominal pain, nausea and vomiting, elevated serum transaminases, and elevated plasma lactate levels. It is related to combination of high blood glucose levels (which promote the flow of glucose into hepatocytes) and exogenous hyperinsulinemia (which stimulates the conversion of glucose to glycogen) [7,8].

The mechanism of growth failure in Mauriac syndrome encompasses insufficient tissue energy (glucose) availability, decreased circulating IGF-1 and a relative growth-hormone-resistant state [1]. It is also has been linked to microvascular complications, particularly to diabetic nephropathy [2]

Conclusion

Mauriac syndrome is still labeled as a rare complication of poor glycemic control in patients with DM type I, yet there is no accurate data of the actual number of cases with Mauriac syndrome worldwide.

Clinical manifestations of Mauriac syndrome of stunted growth, delayed puberty and hepatomegaly are occasionally seen in early adolescent patients. Approach of treatment is continuous insulin therapy and continuous glucose measurements. However, result of catch up growth might be reached but the therapy will not prevent further complications of retinopathy and nephropathy.

Therefore, a tight glycemic control is the golden therapy to delay DM I multisystem complications and Mauriac Syndrome manifestations can be reversed with tight sugar control, adherence to medications and follow ups.

Consent

Written consent was taken from the parents for approval of case reporting and publishing.

Acknowledgment

The authors thank Head of department of pediatrics, scientific committee and academic affairs for granting permission to publish this case.

Conflicts of Interest

None.

Funding

None.

Contribution of Authors

Preparation of first draft: I.M, F.B, F.A, Z.A.

Literature review: F.B, A.D, F.A, Z.A.

Conceptualization: F.B, A.D, F.A, Z.A.

Intellectual inputs for improvement of manuscript: I.M, F.B, N.Z.

Approval of final draft: I.M, N.Z, Z.A.

Bibliography

1. Kocova M and Milenkova L. "Old syndrome-new approach: Mauriac syndrome treated with continuous insulin delivery". *SAGE Open Medical Case Reports* (2018).
2. Mitchell DM. "Growth in patients with type 1 diabetes". *Current Opinion in Endocrinology, Diabetes, and Obesity* 24.1 (2017): 67.
3. MacDonald MJ., *et al.* "Discovery of a genetic metabolic cause for Mauriac syndrome in type 1 diabetes". *Diabetes* 65.7 (2016): 2051-2059.
4. Mayer-Davis EJ., *et al.* "ISPAD Clinical Practice Consensus Guidelines 2018: Definition, epidemiology, and classification of diabetes in children and adolescents". *Pediatric Diabetes* 19 (2018): 719.
5. DiMeglio LA., *et al.* "ISPAD Clinical Practice Consensus Guidelines 2018: Glycemic control targets and glucose monitoring for children adolescents, and young adults with diabetes". *Pediatr Diabetes* 19.27 (2018): 105-114.
6. Vijay Y., *et al.* "Mauriac syndrome: A preventable complication of type 1 diabetes mellitus". *Journal of Obesity and Metabolic Research* 1.4 (2014): 247.

7. Deemer KS and Alvarez GF. "A rare case of persistent lactic acidosis in the ICU: glycogenic hepatopathy and Mauriac Syndrome". *Case Reports in Critical Care* (2016).

8. Pinto MJ., *et al.* "Mauriac Syndrome: A Rare Complication of Type 1 Diabetes Mellitus". *European Journal of Case Reports in Internal Medicine* 5.12 (2018): 764-765.

Volume 9 Issue 11 November 2020