National Board of Examinations - Journal of Medical Sciences
Volume 2, Issue 9, Pages 950–954, September 2024
DOI 10.61770/NBEJMS.2024.v02.i09.013

CASE REPORT

Short Stature with Type-1 Diabetes: A Clinically Observed Case in Patients Suffering From Mauriac Syndrome

Devendrappa K R,[1,*] Nidhi C,[2] Sharique Ahmed,[2] Prithi Jadhav[2] and Shayeebulla Khan[2]

[1]*Senior consultant and DNB PG teacher, Jayanagar General Hospital, Bangalore*
[2]*DNB Medicine PG, Jayanagar General Hospital, Bangalore*

Accepted: 11-August-2024 / Published Online: 08-September-2024

Abstract
Mauriac syndrome (MS) is an exceptionally rare disorder occurring in poorly controlled Type 1 diabetic patients. The consequences include dwarfism, obesity, hepatomegaly, delayed puberty, growth failure and higher levels of transaminase enzyme. We report a case of an adolescent female with classical features of Mauriac syndrome.

Keywords: Growth Failure, Tanner Staging, Over- insulinization, Cushing Syndrome, diabetic cheiroarthropathy, Glucose Monitoring

*Corresponding Author: Devendrappa K R
Email: drdevkowri@gmail.com

Introduction

The global survey claim that the present population are at higher risk of developing diabetes and its related complications. MS is one such rare disorder with a silent feature of glycogenic hepatopathy leading to poor control over type 1 diabetes mellitus (DM) [1]. The patients will have features such as growth retardation (growth failure), obese conditions, hepatomegaly and cushingoid facies. Furthermore, a delayed pubertal menstruation, dyslipidaemia, elevated transaminases and proximal muscle wasting were also observed in such patients [2].

Such type of cases mainly occurs in female and in adolescence, but reports also suggested that the relatively higher cases are observed in children of younger age [3]. All these features can be reversed with the use of insulin injection therapy, instigating prompt control over blood glucose fluxes [4], diet and regular physical exercise.

In our study, one such clinical feature of Mauriac syndrome was observed and the data recorded in a female patient of 18 years age group.

Case Report

This is a case report of an 18 year old female having poorly controlled T1DM from almost one decade. The patient had poorly controlled blood glucose levels and history of growth failure. Our detailed investigations ruled out the history of DKA and the inheritance of diabetes from her family.

The child was subjected to physical examination, with the following observations like short statured height of 132cm (falling below 3rd percentile) and the body weight of 57 kgs. On continuation, the investigational findings depicted BMI of ~32.71, hepatomegaly, Tanner stage I (pre-pubertal) pubertal delay. Along with these features patient was found to have cushingoid face, protuberant abdomen, proximal myopathy, diabetic cheiroarthropathy and scleroderma. On fundus examination, mild to moderate non proliferative diabetic retinopathy was observed.

Laboratory reports revealed the following information: Haemoglobin-(10.4g/dl), fasting blood glucose (FBS-300mg/dl), post prandial blood glucose (PPBS-400 mg/dl), total cholesterol (200 mg/dl), LDL cholesterol (150 mg/dl), HDL cholesterol (23 mg/dl), VLDL cholesterol (50 mg/dl) and triglycerides (160 mg/dl). Likewise, the liver function test depicted total protein, albumin, aspartate transaminase, alanine transaminase, alkaline phosphatase values of 6.4 g/dl, 3 g/dl, 130 U/L, 110 U/L, 170 U/dL respectively. Further the key electrolytes analysis such as calcium, serum sodium, potassium and chloride was found to be 8.1 mg/dl, 130 mmol/L, 4.75 mmol/L, 96.4 mmol/L respectively. The ketone bodies was found to be nil in the urine sample. The Haemoglobin A1C (HbA1C) was as high as 15 mg/dl indicating poorly controlled diabetes.

Table 1.

Hormone Profile	Values
Vitamin D	5.05ng/ml
FSH	4.8μIU/ml
LH	1.8μIU/ml
T3,T4,TSH	Normal

The ultrasound (USG) scan showed evidence of fatty liver and a hypoplastic uterus. The bone age of 14 was confirmed by executing X-ray studies of both the wrist and long bones.

Immediately after diagnosis and laboratory analysis reports, the patient was infused with basal bolus insulin to lower the elevated sugars levels. Over the period of 4 months of monitoring FBS, PPBS and HbA1C were brought up to ~120 mg/dl, 186 mg/dl and 8%, respectively.

Discussion

In the year 1930 a French clinician by name Pierre Mauriac identified Mauriac Syndrome in a 10 year old girl having Type-1 DM with characteristic growth failure and delayed puberty [4]. The MS patient manifests to have some unexpected symptoms closely relating to obesity, hepatomegaly, Cushingoid face, elevated serum liver enzymes, growth retardation, pubertal delay with warning signs of DM.

The pathophysiologic findings primarily suggested that glycogenic hepatopathy, hyperglycaemia and over-insulinization were the reason for liver enlargement and it's impaired functioning [5].

The precise cause of delayed growth is considered to be multifactorial, occurring as one of the subtle features of MS. The cardinal reason could be low glucose tissue metabolism, altered hormone receptor action, hypercortisolism, decreased GH and IGF-1 [1,6].

Genetics of MS

It can be said that a person suffering from MS has increasing incidence to develop type-1 DM leading to insulin deficiency, in-turn high sugar levels. But on the other hand there is no evidence of a diabetic patient having risked with Mauriac conditions. This rules out the possibility that elevated glucose levels may not be the potential reason for developing MS.

An adolescent boy was detected to have severe MS linked with mutation of exon 9 of Phosphorylase kinase (PHK) G_2 gene, which is the catalytic subunit of the enzyme Glycogen Phosphorylase kinase (PhK), the first enzyme in the pathway of glycogen metabolism [7].

Moon face is often associated with Cushing's syndrome mainly resulting due to glycogen deposition (with pituitary adrenal axis intact).

Liver biopsy in the setting of MS demonstrates steatosis and glycogen deposition, though the findings can vary in presentation [8]. Hyperglycaemia and Low insulin levels caused by poor T1DM leads to fatty acid deposition in the liver that roots to hepatomegaly and characteristic liver biopsy identifications. By controlling the overall glycaemic conditions in an individual these findings could be reversed [9].

A group of clinicians lead by Mauriac concluded a Nil report related to hypothalamic pituitary dysfunction with respect to growth failure [10] and growth failure regresses with adequate insulinization in patients with MS. However, on continuation of treatment with insulin further resulted in deterioration of retinopathy and nephropathy [11].

Pubertal issue leading to menstruation delay in MS can be normalized by controlled insulin therapy. For instance, a clinical team lead by Traisman were able to demonstrate successful sexual development and pregnancies in a 22 years old MS Patient [12].

Conclusion

MS is a rare complication emphasizing the poorly controlled DM in adolescence aged population. The unlikely causes of the MS could be reversed by prompt insulin dosing and timely glucose monitoring. This could be an ideal approach to treat the subject leading to high indexed clinical outcome with lesser life-threating complications.

Ethics Declarations

Funding

This study did not receive any funding.

Conflict of Interest

The authors declare that they have no competing interests.

Ethics approval, Consent to participate, Consent to publish, Availability of data and material, Code availability

Not applicable.

References

1. Mahesh S, Karp RJ, Castells S, Quintos JBQ. Mauriac syndrome in a 3-year-old boy. Endocr Pract. 2007;13(1):63-6.
2. Gutch M, Philip R, Saran S, Tyagi R, Gupta KK. Re-emergence of a rare syndrome: A case of mauriac syndrome. Indian J Endocrinol Metab. 2013;17(1):S283-5.
3. Madhu SV, Jain R, Kant S, Prakash V. Mauriac syndrome: A rare complication of type 1 diabetes mellitus. Indian J Endocrinol Metab. 2013;17(4):764-5.
4. Kim MS, Quintos JB. Mauriac syndrome : Growth failure and type 1 diabetes mellitus. Pediatric Endocrinol Rev. 2008;5:989-93.
5. Rogal SS, Ukomadu C, Levy BD, Loscalzo J. Clinical problem-solving. A sweet source of abdominal pain. N Engl J Med. 2011;364(18):1762-7.
6. Lee RGL, Bode HH. Stunted growth and hepatomegaly in diabetes mellitus. J Pediatr. 1977;91(1):82-4.
7. MacDonald MJ, Hasan NM, Ansari I-UH, Longacre MJ, Kendrick MA, Stoker SW. Discovery of a genetic metabolic cause for Mauriac syndrome in type 1 diabetes. Diabetes. 2016;65(7):2051-9.
8. Lorenz G. Bioptical liver changes in Mauriac syndrome. Zentralbl Allg Pathol. 1981;125(4):364-8.
9. Farrell M, Bucuvalas J. Systemic disease and the liver. In: Ch, Philadelphia L, Williams W, editors. Liver disease in children. 2001;897-927.
10. Mauras N, Merimee T, Rogol AD. Function of the growth hormone insulin-ike growth factor 1 axis in the profoundly growth-retarded diabetic

child: evidence for defective target organ responsiveness in the Mauriac syndrome. Metabolism. 1991;4010:1106-11.

11. Kim MS, Quintos JB. Mauriac syndrome: growth failure and type 1 diabetes mellitus. Pediatric Endocrinology Rev. 2008;5 Suppl 4:989–93.
12. Traisman HS, Traisman ES. Mauriac's syndrome revisited. Eur J Pediatr. 1984;142(4):296–7.