A Case Report of Mauriac Syndrome: Diabetic Dwarfs

Jaydeep A. Patel[1*], B. B. Solanki[2], Rishabh R. Gupta[3], Prashant Dudhagara[4]

[1]3rd Year Resident, [2]Professor & Head of Unit, [3]2nd Year Resident, [4]1st Year Resident, Dept. of Medicine, B.J.Medical College, Ahmedabad.

ABSTRACT

Introduction: Mauriac syndrome is a severe form of growth retardation seen in patients with poorly controlled type 1 diabetes mellitus characterized by growth failure, delayed puberty, hepatomegaly and Cushingoid features. It is often referred to as diabetic dwarfism. The actual cause is unknown but is probably a combination of factors including inadequate glucose in the tissues, decreased IGF-1 and growth hormone levels, impaired bioactivity of the hormones, a circulating hormone inhibitor, resistant or defective hormone receptors, especially with the use of premix insulin. **Case Report:** We report a case of 15 year old female, known case of type 1 diabetes mellitus since the age of 5 years, presented to us with diabetic ketoacidosis. She had multiple similar episodes in the past. She was on premix insulin (30/70) and tablet metformin but had poor compliance to the treatment. On examination she was found to be significantly short for her age (height 129 cm, less than 3rd percentile) and weight was 27 kg (less than 3rd percentile) with cushingoid features (round face and protuberant abdomen). She was at Tanner stage P1. On detailed laboratory evaluation, IGF-1 levels were decreased (64), LH levels were below normal limits (0.2) and FSH levels were normal, while transaminases and HbA1C (8.3) were elevated. TSH was elevated (22.6), anti-TPO was elevated (252.6) with normal FT3, FT4. Hemogram, renal function test, GH, cortisol, estradiol, prolactin, S. iron, S. ferritin were normal. Ultrasound abdomen showed fatty liver with atrophic pancreas and USG neck showed bilateral bulky thyroid lobes. Fundus examination was normal. Now for the past 1 year she is on regular insulin 8/15/8 units (BBF/BL/BD) & Insulin Glargine 12 units. This led to a decrease in hepatomegaly and increase in height of 2 cms. **Conclusion:** Mauriac syndrome is a rare complication of poorly controlled diabetes mellitus in adolescence, but the treating physician should keep a high index of suspicion for this so that proper growth can be accomplished with timely intervention.

Key words: Mauriac syndrome

Introduction

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* **Corresponding Author:**
Dr. Jaydeep A. Patel
Email: dr.jaydeep88@gmail.com

glucose in the tissues, decreased IGF-1 and growth hormone levels, impaired bioactivity of the hormones, a circulating hormone inhibitor, resistant or defective hormone receptors, especially with the use of premix insulin.

Case Report

We report a case of 15 year old female, known case of type 1 diabetes mellitus since the age of 5 years, presented to us with diabetic ketoacidosis. She had multiple similar episodes in the past. She was on premix insulin (30/70) and tablet metformin but had poor compliance to the treatment.

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Discussion

Mauriac syndrome, first described in 1947, is a rare complication associated with T1DM and is typically associated with poor insulin compliance and glycemic control. The features of Mauriac syndrome are mostly related with deficient insulinization. Hepatomegaly can develop in patients due to intrahepatic glycogen deposition; if these patients also have elevated liver enzymes, cushingoid features, dyslipidemia and delayed growth maturation[1], Mauriac syndrome can be diagnosed. Two different forms of Mauriac syndrome was defined by the presence of obesity. The first form which is treated with regular insulin alone is associated with cushingoid obesity with secondary hyperadrenalism. Mauriac syndrome has been reported in patients who are not obese and it occurs in patients who have been given regular, under the dose insulin. Although hepatomegaly and elevated transaminases are common findings in Mauriac syndrome, other described pediatric manifestations can include malnutrition, growth failure and development of cushingoid features. Malnutrition associated with poor T1DM control also can lead to false elevation of the sweat chloride level, so such patients can present with false positive screens for cystic fibrosis. Cases of Mauriac syndrome show that normal insulinization and normal caloric intake are factors that affect the synthesis and secretion of IGF-1. Low level of IGF-1 secretion in our patient was explained by a low dose insulin over a long time, and delayed puberty. Insulin and sex steroids have a stimulatory effects on the synthesis and secretion of IGF-1. This hypothesis was supported by the fact that the IGF-1 levels of our patient were in normal

ranges after the normalization of caloric and insulin intake[2]. T1DM is associated with other autoimmune diseases, including celiac disease and autoimmune thyroiditis[3], and it is common for patients with T1DM to have elevated autoantibody titres. In this case, a 15 year old female with poorly controlled T1DM presented with elevated serum transaminases. A subsequent liver biopsy was consistent with Mauriac syndrome, which demonstrated steatosis as well as glycogen deposition[4]. Poorly controlled T1DM leads to fatty acid transport to liver, due to hyperglycemia and low insulin levels, which causes hepatomegaly and characteristic liver biopsy findings. These findings reversed with improved glycemic control[5]. A subsequent liver biopsy demonstrated normal portal tracts; however, hepatocytes demonstrated cytoplasmic clearing secondary to increased intracellular glycogen and microvescicular fat. This biopsy was consistent with Mauriac syndrome and the importance of improved adherence to insulin therapy was explained to the patient and her family.

Conclusion

Mauriac syndrome is a rare complication of poorly controlled diabetes mellitus in adolescence, but the treating physician should keep a high index of suspicion for this so that proper growth can be accomplished with timely intervention.

References

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