Case Report

Mauriac Syndrome: An Infrequent Encounter in Type 1 Diabetes Mellitus

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Abstract

Mauriac syndrome is a rare complication of poorly controlled type 1 diabetes mellitus. It is characterised by poor glycemic control, growth impairment, delayed puberty, cataract, retinopathy, hepatomegaly, nephropathy and cushingoid features such as moon facies, protruded abdomen and muscle wasting.

We hereby report a case of Mauriac syndrome in an eight year old female child.

Keywords: Mauriac Syndrome; Type 1 Diabetes Mellitus; Rare Complication; Poor Glycemic Control

1. Introduction

Type 1 Diabetes Mellitus is one of the common endocrinological morbidity in pediatric population and its various complications in this population are not unknown. Mauriac syndrome is one of such complications characterised by poor glycemic control, growth impairment, delayed puberty, cataract, retinopathy, hepatomegaly, nephropathy, cushingoid features such as moon facies, protruded abdomen and muscle wasting [1-3].

This syndrome has become quite a rare entity now-a-

days, especially in developed countries owing to introduction of long acting insulin, better compliance and good glycemic control. Very few cases have been reported in India [4, 5]. Therefore, we hereby report a case of Mauriac syndrome.

2. Case Report

An eight year female child was referred to us as a known case of type 1 diabetes mellitus for further management in view of poorly controlled blood sugar levels. On eliciting further history, the child complained of diminished vision since one month. The child was diagnosed with type 1 diabetes mellitus at the age of 7 years following which the child was started on subcutaneous mixed insulin preparation containing prolonged-acting and short-acting human insulin.

However, the treatment compliance was not good because there was improper injection technique, poor adherence to injection schedule as well as poor blood sugar monitoring. On examination, the child was conscious, cooperative, well oriented to time, place and person. She had short stature (109 cm, less then 3rd percentile) and was underweight (17.74 kg, less then 3rd percentile). Her BMI was 14.9 kg/ m2 (between 5^{th} and 10th percentile). Her Sexual Maturity Rating (SMR) stage was 1. She had cataract of both eyes. Her abdomen was protruded with hepatomegaly.

On investigating, her complete hemogram, liver and renal function tests, serum electrolytes and blood gas parameters were found to be normal. Urine study showed ketonuria, non-nephrotic range proteinuria (spot urine microalbumin ratio 0.46) and glycosuria. Her fasting blood sugar was 487 mg% with HBA_1C (glycosylated haemoglobin) was 11.7% (normal < 6%). Her thyroid function was within normal limit. Her IA2 (Islet Antigen 2) antibody and GAD 65 (Glutamic Acid Decarboxylase 65) antibody titres were positive. Ultrasound abdomen revealed hepatomegaly with fatty infiltration of the liver. Her clinical history, examination and investigation findings, in the background of type 1 diabetes mellitus, pointed to the diagnosis of Mauriac syndrome.

Strict diabetic dietary management was started along with regular monitoring of blood sugar level. Long acting insulin Glargine injection was administered once daily along with regular insulin injection before breakfast, lunch and dinner after titration as per blood sugar level (basal-bolus regimen). Supplements such as calcium, vitamin D3 and multivitamin were added. The child followed up after 2 months of therapy with well controlled blood sugar level and reduction in liver size both clinically and on ultrasound. She also underwent cataract surgery in both eyes and subsequently had improved vision.

3. Discussion

Pierre Mauriac in 1930 described this syndrome in children diagnosed as type 1 diabetes mellitus who presented with growth failure, pubertal delay, abdominal distension and hepatomegaly and who were treated with short-acting insulin [6]. Its incidence is equal in male and female population with adolescence being the most affected group [7]. Its etiology is multifactorial such as decreased levels of insulin-like growth factor-1 (IGF-1) and growth hormone, defective or resistant hormone receptors or inadequate utilisation of glucose in the tissues [8]. Hepatomegaly is thought to be due to glycogen deposition in the liver [9, 10].

Recently, a genetic cause for metabolic abnormality in Mauriac syndrome has been discovered in the form of

mutation in the catalytic subunit of liver glycogen phosphorylase kinase [11]. Liver glycogen phosphorylase kinase normally activates glycogen phosphorylase which catalyses the first step in glycogen breakdown. The mutation acts in a dominant fashion to completely inhibit glycogen phosphorylase kinase activity resulting in glycogen deposition in the liver cells [11]. Many clinical features such as growth impairment, pubertal delay and hepatomegaly revert with improved glycemic control [4]. Continuous insulin delivery through insulin pump may be a useful treatment approach to combat the clinical features of Mauriac Syndrome [8, 12]. With this approach, stepwise metabolic control can be achieved [13].

In our case, the female child had poorly controlled diabetes mellitus with short stature, cataract, protruding abdomen with hepatomegaly which led us to the diagnosis of Mauriac syndrome. The causative factor in this case attributed to poor compliance of the patient and her family in form of improper injection technique, poor adherence to injection schedule as well as poor blood sugar monitoring. This poor compliance, in turn, is attributed to the ignorance of the family about the disease and its consequences.

Conflict of Interest

We, the authors declare that there is no conflict of interest.

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