

MAURIAC SYNDROME WITH CELIAC DISEASE: RARE ASSOCIATION

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Abstract: - *Growth failure in type 1 Diabetes Mellitus can occur for several reasons. Mauriac syndrome is a rare cause of severe growth failure in type 1 Diabetes Mellitus. The classical severe presentation of Celiac Disease rarely occurs in type 1 Diabetes Mellitus patients, but more often patients have few/mild symptoms of Celiac Disease or are completely asymptomatic (silent CD). In fact diagnosis of Celiac Disease is regularly performed by means of the screening in type 1 Diabetes Mellitus patients. There may be different forms and etiologies involved in Mauriac syndrome. We hereby report a case of Mauriac syndrome in type 1 Diabetes Mellitus with simultaneous celiac disease which is very rarely reported.*

Keywords: *Mauriac syndrome, Celiac Disease, Type 1 Diabetes Mellitus*

Introduction:

The association of dwarfism, obesity and hepatomegaly in patients with insulin-dependent diabetes mellitus (T1DM) is known as Mauriac syndrome. Its presenting features include obesity, hepatomegaly, elevated transaminase levels and cushingoid facies [1], and is generally associated with poor glycemic control in adolescents. The physician should have a high index of suspicion regarding the development of Mauriac syndrome if uncontrolled T1DM is associated with growth failure and delayed puberty, since these changes are reversible with proper glycemic control. [2] Celiac Disease (CD) occurs in patients with Type 1 Diabetes Mellitus ranging the prevalence of 4.4-11.1% versus 0.5% of the

general population. The mechanism of association of these two diseases involves a shared genetic background: HLA genotype DR3-DQ2 and DR4-DQ8 are strongly associated with type 1 Diabetes Mellitus, DR3-DQ2 with Celiac Disease. [9]

Case History:

A 14-year-old male with a 5 years history of T1DM was admitted for evaluation of growth retardation, abdominal distension and diabetes. He had uncontrolled blood sugar levels despite being compliant on pre-mix insulin (30/70) injection in two divided daily doses of 15 units before breakfast and 10 units before dinner. He had landed up into multiple episodes of documented hypoglycaemia while being on the above regimen. However, there

was no history suggestive of any episode of diabetic ketoacidosis, and none of his family members were diabetic.

Physical Examination:

Anthropometric data revealed height 120 cm (<3rd percentile), weight 21 kg (<3rd percentile), body mass index 14.58 kg/m², abdominal circumference of 65 cm, MAC of right arm 13 cm and left arm 12.5 cm, liver span of 18 cm. BP 90/60 mmHg right arm supine position.

He had typical cushingoid “moon-like” facies, with thinning of scalp hair, cold extremities and low pitched voice. His abdomen was protuberant, and hepatomegaly was elicited on physical examination. Sexual Maturity Rating (SMR) revealed genital and pubic hair development at “tanner stage 1” for both, i.e. G1 and P1 respectively. Testicular volume was pre-pubertal (<2ml) as compared with a Prader orchidometer. There was no vitiligo or Hyperpigmentation.

Laboratory Analysis:

Haemoglobin 7.7 g/dl, total protein 7.65 g/dl, albumin 4.20 g/dl, triglycerides 391 mg/dl, total cholesterol 122 mg/dl, High density Lipoprotein-cholesterol 21.2 mg/dl and Low density Lipoprotein-cholesterol 47.7 mg/dl, aspartate aminotransferase 70.6 U/L (5-45 U/L), alanine aminotransferase 76.8 U/L (5-45 U/L) and ALP 283 U/L. Serum calcium was 10.2 mg/dl (8.7-10.2mg/dl) and phosphorus was 5.6 mg/dl (2.5-4.3 mg/dl). Blood urea was 14.6 mg/dl and serum creatinine was 0.55 mg/dl. Serum sodium was 138mEq/l and potassium was 3.75mEq/l and serum osmolality was 275 mOsm/kg (275-295 mOsm/kg). His 24 h urinary protein excretion was 1512 mg (< 150mg/day). His thyroid

profile T3-1.09, T4-29.15 and TSH-3.52, HbA1c = 4.46%. Ultrasound study of abdomen showed the presence of fatty infiltration of liver (Grade I) with hepatomegaly. Serum tissue transglutaminase IgA was more than 300 U/MI (normal < 12). OGD revealed flattened duodenal mucosal fold and biopsy revealed villous atrophy.



Figure a and b: Type 1 diabetes with prominent abdomen, short stature and celiac disease

Our patient presented with uncontrolled type 1 diabetes mellitus, delayed puberty, severe growth retardation and hepatomegaly, thus enabling a diagnosis of Mauriac syndrome to be made. Upon investigation patient was found to have celiac disease. Few cases have been described of Mauriac syndrome with simultaneous celiac disease

Discussion:

Mauriac syndrome, the association of dwarfism, obesity and hepatomegaly in patients with Insulin Dependent Diabetes Mellitus (T1DM), occurs most commonly during adolescence, has no sex predilection and has an uncertain aetiology. Most likely a combination of factors comes into play, including inadequate glucose utilization by the tissues, lowered growth hormone and insulin-

like growth factor-1 (IGF-1) levels, reduced bioactivity of hormones, circulating hormone inhibitors, defective receptor signalling, insulin deficiency leading to poor glycemic control, decreased caloric intake and concurrent Autoimmune diseases. The most commonly associated autoimmune diseases include Addison's disease, hypothyroidism, celiac disease and autoimmune gastritis [3].

In children having DM and growth problem may mean they also have celiac disease. [4] Conversely, recurrent hypoglycaemia can be a sign of celiac disease in people with type 1 DM [5]. Previously, when long-acting insulin was unavailable, delays in growth and puberty were common; with the availability of better treatment modalities, this syndrome is a rare occurrence in the present times. Owing to the frequent absence of symptoms suggestive for gluten-sensitive enteropathy, serological screening for CD acquires much more importance in T1DM than in other patient settings. The diagnostic accuracy of serological tests for identifying CD in T1DM is very high, but some pitfalls should be considered in the evaluation of antibody tests. tTGA is the first choice test owing to their higher sensitivity for CD (~98%), but these markers give a number of false positives at T1DM onset. Duodenal histology remains the gold standard for CD diagnosis in T1DM. The advantages of GFD in T1DM patients with CD are evident with improvement of intestinal and extra intestinal CD-related symptoms, whereas the course of T1DM is not influenced so much by gluten withdrawal. Indeed, it has been demonstrated that hypoglycemic episodes decrease, but at the same time insulin needs increase and HbA1c levels do not improve. Moreover, the protection induced by GFD on the development of lymphoma and other complications in CD patients overcomes some negative effects of

the diet, such as the possible development of a metabolic syndrome, which is favoured by the improved intestinal absorption and by a high consumption of commercial gluten-free foods, particularly rich in carbohydrates and lipids.

Conclusion:

Through Mauriac's syndrome nowadays found to be rare in developed countries, is still prevalent in developing countries. Timely identification of this syndrome can prevent various complications associated with this syndrome.

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