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Two Cases of Testicular Adrenal Rest Tumor (TART)

Hatice Özışık[1], Banu Şarer Yürekli[1], Ilgın Yıldırım Şimşir[1], İlker Altun[1], Utku Soyaltın[1], Ezgi Güler[2], Hüseyin Onay[3], Banu Sarsık Kumbaracı[4], Füsun Saygılı[1]

[1]Ege University Faculty of Medicine, Department of Endocrinology and Metabolism Diseases, İzmir, Turkey

[2]Ege University Faculty of Medicine, Department of Radiology, İzmir, Turkey

[3]Ege University Faculty of Medicine, Department of Medical Genetics, İzmir, Turkey

[4]Ege University Faculty of Medicine, Department of Pathology, İzmir, Turkey

Testicular adrenal rest tumor (TART) is a benign tumor which is seen in male patients who have congenital adrenal hyperplasia (CAH).

Case 1: A 24-year-old male was diagnosed with CAH. He took hydrocortisone treatment upto 18 years of age however discontinued it thereafter. Testicular mass was detected and right radical orchiectomy was performed; testicular tumor of adrenogenital syndrome was determined. On physical examination, height was 152 cm, weight was 47 kg, BMI was 20.3 kg/m^2. He had short fingers. In laboratory examination, 17-OHP 122 ng/mL (0.6-3.3), adrenocorticotropic hormone 118 pg/mL (< 46), free testosterone 34.4 pg/mL (57-178), DHEAS 378.4 µg/dL (85-690), and cortisol 5.2 µg/dL were detected. In CAH mutation screening, mutations in an allele (heterozygous) I2 splice and in the other allele (heterozygous) L307 frameshift were detected. Dexamethasone 0.75 mg once daily was initiated.

Case 2: A 38-year-old male has followed with diagnosis of Addison disease for 35 years. Right testicular tumor was defined as Leyding cell tumor in 2010. In scrotal USG, small multifocal lesions were detected and testicular biopsy was done which revealed testicular tumor of adrenogenital syndrome. He took 30 mg hydrocortisone once daily. On physical examination, height was 174 cm, weight 104 kg, and BMI was 34.4 kg/m^2. In laboratory examination, 17-OHP 157 ng/mL (0.6-3.3), adrenocorticotropic hormone 194 pg/mL (< 46), free testosterone 31.6 pg/mL (57-178), DHEAS 123.5 µg/dL (85-690), and cortisol 2.14 µg/dL were detected.

TARTs are usually seen bilaterally (83%) and histopathologically it is difficult to differentiate them from Leydig cell tumor. It should be kept in mind that testicular USG is of significant importance in early diagnosis of TART.

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Hepatic Glycogenosis in a Patient with Type 1 Diabetes: Mauriac Syndrome vs. Congenital Glycogen Storage Disease

Ömercan Topaloğlu[1], Sibel Demiral Sezer[2], Bilgin Demir[3], Abdullah Serkan Yener[4]

[1]İnönü University Faculty of Medicine, Department of Endocrinology, Malatya, Turkey

[2]İzmir Tepecik Training and Research Hospital, Clinic of Internal Medicine, İzmir, Turkey

[3]Recep Tayyip Erdoğan University Faculty of Medicine Training and Research Hospital, Clinic of Internal Medicine, Rize, Turkey

[4]Dokuz Eylül University Faculty of Medicine, Department of Endocrinology, İzmir, Turkey

We aimed to emphasize that the etiology of elevated liver enzymes should clearly be explained in type 1 diabetes mellitus patients.

A 20-year-old male who was on intensive insulin regimen with a history of type 1 diabetes for 11 years was admitted to our hospital with complaints of diarrhea and vomiting for three days. No other chronic diseases or complications of diabetes were present. Physical examination revealed temperature of 37.6 °C, pulse 138/min, blood pressure 86/40 mmHg, body weight 45 kg, and height of 155 cm, as well as Cushingoid face features and hepatomegaly. Examination of the other systems was unremarkable.

Laboratory findings were as followings: Venous plasma glucose 348 mg/dL, urine ketone 2+, and metabolic acidosis. The patient was managed by diabetic ketoacidosis protocol. The patient had also elevated ALT (167 U/L), AST (184 U/L), GGT (180 U/L), and ALP (140 U/L) levels. Abdominal ultrasonography performed to figure out the elevated liver enzymes showed grade 1 hepatosteatosis and hepatomegaly. Due to history of poorly controlled diabetes, hepatic glycogen deposition was also considered. Liver biopsy demonstrated PAS(+) granules in hepatocytes. We diagnosed the patient as Mauriac syndrome with the findings of growth retardation, poorly controlled diabetes, hepatomegaly, and hepatic glycogenosis. Genetic analysis was performed to exclude congenital glycogen storage disease type-1 (GSD-1). Heterozygous mutation was found in glucose-6-phosphatase (17q21,p.R83C), describing our patient as carrier. Although it is known that the carriers are asymptomatic, we assume that this mutation could contribute to hepatic glycogenosis. Diet and medical therapy were planned.

Hepatomegaly and elevated transaminases in type 1 diabetes patients may be caused by hepatic glycogenosis. In differential diagnosis of Mauriac syndrome, congenital glycogenoses should also be considered.