

17 HYDROXYLASE DEFICIENCY AND DIABETES IN A CASE WITH 46 XY FEMALE

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INTRODUCTION

In this presentation, we have a interesting patient in female appearance with puberty delay at the age of 13. The patient had a 46 XY karyotype.

The patient was diagnosed with 17 hydroxylase deficiency. Hydrocortisone treatment was started and bilateral orchioectomy was performed. Later, type 2 diabetes developed.

AIM

In this case, we wanted to emphasize that 17 hydroxylase deficiency, which is rarely seen, is a cause of gender development disorder and to discuss the surprising developed diabetes mellitus in the follow-up.

METHOD

This presentation is case report.

RESULTS

The patient was externally in female appearance and there was no puberty development. Labia minora was seen. The urethral vaginal openings were in the vestibulum and bilateral gonads could not be palpated in the inguinal canal.

Uterus and ovaries were not observed in USG.

There was hypergonadotropic hypogonadism and high level of progesterone with adrenal insufficiency; ACTH 115 ng /L (7.2-63), cortisol 0.43 µg / dL, progesterone 8.45 µg / L (0.31-1.52). Hydrocortisone treatment with 20 mg/m²/day dose to the patient was started.

In the genetic study, CYP 17A1 (NM_000102.3) mutation was detected in both alleles (homozygous) c.1319G> (p.Arg440His).

The patient was consulted with Pediatric Surgery. Bilateral orchioectomy was performed by laparoscopy.

After the operation, estradiol 0.5 mg orally was started. In the follow-up, high level of blood sugar was observed 3 weeks after the operation. HbA1C was 8.5%, C-peptide 3.51 µg / L (0.9-7.1), insulin 13.05 mU / L (1.9-23), diabetes autoantibodies (-) in laboratory.

CONCLUSIONS

17 hydroxylase deficiency should also be considered in patients presenting with delayed puberty.

Gonadectomy should be performed to prevent tumor development.

Since hydrocortisone is used in the treatment of patients, diabetes may develop in people with a genetic predisposition.

Patients should also be followed up for signs of diabetes.

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