



# 17 ALPHA HYDROXYLASE, 17-20 LYASE DEFICIENCY, A CASE WITH HYPOCALCEMIC SYMPTOMS

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## BACKGROUND

17 alpha hydroxylase, 17-20 lyase deficiency is an autosomal recessive inherited congenital adrenal hyperplasia type which is due to mutation in CYP17A1 gene and characterized with adrenal and gonadal sex steroid deficiency, delayed puberty in girls, XY sex development disorder in boys and hypergonadotrophic hypogonadism in both gender.

## CASE

15 year old girl referred to our clinic with vomiting, fatigue and muscle spasms. In her physical examination weight was 25-50 centile, height was 75-90 centile, The patient's blood pressure was 120/70 mmHg, Her breast development was at Tanner stage 1. The bone age was determined 11 years. There was no axillary or pubic hair. The patients laboratory results were glucose: 98 mg/dL K: 2.1 mEq/L, Na: 142 mEq/L, Ca: 9,3 mg/dl , iCa: 0,94 mmol/L (1,16-1,34 mmol/L), FSH: 59,2 mIU/mL , LH: 64,4 mIU/, E2: <5 pg/mL , total testosterone:<0,025ng/dl, cortisol: 4,59 µg/dL, ACTH: 129 pg/ml, renin: 2,05 ng/mL/hour (0.98—4.18), aldosterone: 11,01 ng/dL (4-48 ng/dL), 17 OH progesterone :0,43 ng/ml , DHEA-S :1,4 µg/dL, TSH: 0,61 µIU/mL, free T4: 1.49 ng/dL , prolactin: 13,79 ng/ml, progesterone: 2,79 ng/ml , 11 deoksikortisol : 0,81 ng/ml), the blood gases examination results were ph: 7.60, HCO<sub>3</sub>: 37,3 mmol/L, PCO<sub>2</sub>: 57,1 mmHg, and base excess: 14. In the suprapubic ultrasonography of the patient, there were no mullerian or wolfian duct. Karyotype was 46 XY and SRY (+).Treatment with hydrocortisone at a dose of 15 mg/m<sup>2</sup>/day was started. Hormone replacement therapy and laparoscopic gonad examination was planned. We are waitig for genetic

examination result about 17 alpha hydroxylase, 17-20 lyase deficiency.

## DISCUSSION

17 alpha hydroxylase, 17-20 lyase deficiency is a rare congenital adrenal hyperplasia type which presents with hypertension, hypokalemia and hypogonadism. But this case was referred our clinic with hypocalcemic symptoms due to metabolic alkalosis.



\*There is no conflict of interest

