Novel CYP17A1 mutation and CYP21 mutations in two siblings

Emregül Işık [1], Mehmet Keskin[2], Ahmet Yeşilyurt[3]

[1]Pediatric endocrinology, Gaziantep Children's Hospital, [2] Pediatric endocrinology, Gaziantep University Faculty of Medicine,[3] Genetics,Yıldırım Beyazıt Education and Training Hospital, Ankara

Introduction

17 hydroxylase deficiency is a rare form of congenital adrenal hyperplasia resulting from loss-of-function mutations involving the *CYP17* gene. It is characterized by decreased production of glucocorticoids and sex steroids and increased synthesis of mineralocorticoid precursors, resulting in varying degrees of hypertension and hypokalemia.

Clinical Case

14 year old girl

Presentation symptoms:

Absence of breast development, abdominal pain, loss of appetite

History:

No drug usage or any other known chronical illness

Bilateral inguinal hernia operation at 9 years

Parents were first degree cousins

Physical Examination

Weight: 37 kg (-2.8 SDS)

Height: 152.5 cm (-1.3 SDS)

BMI: 15.9 (-1.5 SDS)

Puberty: Tanner stage 1.

Blood pressure was 120/90 mmHg

Laboratory

Na: 140 mmol/l, K: 3.6 mmol/l, glukoz: 95 mg/dl

FSH: 94 ıu/l, LH: 63 ıu/l, Estradiol: 11.8 pg/ml, PRL: 9.3 ng/ml

ACTH: 1250 pg/ml (0-46), cortisol: 0.5 µg/dl

Total testosteron: 10 ng/dl (5-40), DHEASO4: 15 µg/dl (35-430)

Progesteron: 12.7 ng/ml (0-1.24)

pH:7.34, HCO3: 26.4 mmol/l.

TSH: 2.16 mıu/ml, fT4: 1.02 ng/dl (0.8-2)

Imaging

Uterus and ovaries were not visualized.

Karyotype: 46 XY

CYP17A1 Gene Analysis

Novel homozygous c.617_618delTA mutation in exon 3.

Parents and siblings were heterozygous.

Newborn Sibling

Presentation symptom: ambiguous genitalia.

Phallus was 2 cm with no palpable gonads.

Salt wasting at 2 months.

Laboratory

17 hidroksiprogesteron: 215 ng/ml

Total testosteron: 245 ng/dl

1-4 androstenedion >10 ng/ml.

Karyotype: 46 XX.

CYP21 Gene Analysis

Homozygous CYP21 c.293-13A/C>G (In2G) mutation

Results

17 hydroxylase deficiency causes ambiguous genitalia in 46 XY individuals while sexual infantilism is seen in 46 XX patients secondary to decreased production of sex steroids. We present two different types of congenital adrenal hyperplasia, classical 21 hydroxylase deficiency adn 17 hydroxylase deficiency in two siblings which is rare.

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Gonads & DSD

Emregl Isik

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