



A 46, XX PATIENT WITH 21-OHD DIAGNOSED DURING THE ETIOLOGIC WORKUP OF MALE INFERTILITY

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Introduction

Congenital adrenal hyperplasia (CAH) is a group of autosomal recessive disorders of the steroid biosynthesis. 21-hydroxylase deficiency (21-OHD) represents 90–95% of CAH patients and causes adrenal insufficiency and virilization. Although most patients are diagnosed in childhood, the diagnosis of some classical CAH cases are extremely delayed up to sixth and seventh decades of life. Herein, we report a 46, XX patient with 21-OHD diagnosed during the etiologic workup of male infertility.

Table 1. Biochemical characteristics of the patient

FSH (mIU/ml)	0.26
LH (mIU/ml)	<0.1
E ₂ (pg/ml)	72.7
Total Testosteron (ng/dl)	1075
ACTH (pg/ml)	469 (<46)
Cortisol (ng/ml)	35.68 (50-250)
Na (mmol/L)	124
K (mmol/L)	3.8
17-OH Progesteron (ng/ml)	186.89 (0.42-1.96)
DHEA (ng/ml)	6.87 (0-10)
DHEA-S (ng/ml)	1084.63 (650-3340)
Androstenedione (ng/ml)	46.05 (0.4-1.5)
Testosterone (ng/ml)	4.81 (2.4-9.5)
Dihydrotestosterone (ng/ml)	0.21 (0.16-0.79)
Corticosterone (ng/ml)	0.96 (0.53-15.6)
11-deoxycorticosterone (ng/ml)	<0.053 (0-0.15)
11-Deoxycortisol (ng/ml)	0.64 (0.1-0.79)
Aldosterone (ng/ml)	0.04 (0-0.21)
Progesterone (ng/ml)	1.57 (0.2-1.4)
Pregnenolone (ng/ml)	0.78 (0.3-2.08)
17-OH Pregnenolone (ng/ml)	1.88 (0-1.28)

Case Report

A 32-years-old male patient was referred due to infertility. His parents were second degree cousins, he or his family had no history of any chronic disease. He a deceased 4 years old brother of an unknown cause. The physical examination of the patient revealed genital hyperpigmentation, bilateral cryptorchidism, the phallus was approximately 3 cm and had no hypospadias. Ultrasonography revealed a hypoplastic uterus (61x13 mm) but no ovarian tissue. The biochemical data suggested CAH due to 21-OHD (Table 1). His karyotype was 46, XX and SRY was negative. Genetic testing demonstrated homozygous *CYP21A2* A/C-656->G mutation. Steroid replacement and surgical management were planned but the patient refused treatment.

Conclusion

Although classical 21-OHD may cause a life threatening salt-wasting crises in early infancy and childhood, some cases may have atypical delayed presentations. This may potentially be due to compensatory effect of adrenal steroid hormone precursors that accumulate and transactivate glucocorticoid receptor, and mask cortisol deficiency. Detailed genital examination should be a part of diagnostic approach in order to recognise adrenal causes of infertility.

