Late complications in a man with poorly controlled congenital adrenal hyperplasia – case report

K. Maliszewska, A. Popławska- Kita, I. Kowalska, Maria Górska

Department of the Endocrinology, Diabetology and Internal Medicine, Medical University of Białystok,, Poland

Introduction

We report a case of CAH who developed several complications due to poor control. Proper diagnosis and treatment can enable men with the disease to have a normal life and fertility.

Case report

39-year-old patient with a history of primary adrenal insufficiency was admitted to our Department because of infertility. Semen analysis performed prior to hospitalization, showed azoospermia. The patient did not have medical documentation regarding his disease and was not able to explain exactly why the glucocorticoid therapy was initiated. In referral letter to the hospital, Addison's disease was written in the diagnosis. We learned from parents, that hydrocortisone was introduced after life-threatening episode in the neonatal period and maintained untill now. The study examination revealed dark complexion and brown spot on mucous membranes, livid lips, and enlarged testes with palpable nodules. Laboratory studies have shown: increased level of RBC, hyperlipidemia, high values of ACTH (> 1250pg / ml) and testosterone (> 15 ng / ml) and low concentrations of gonadotropins. After two days of hydrocortison withdrawal, cosyntropin test was performed revealing impaired cortisol response. Moreover, basal level of 27-OH progesterone and after cosyntropin stimulation was very high. The abdominal computed tomography scan revealed grossly enlarged and heterogeneous adrenal glands (right 39x71x70mm, left 38x89x110mm). They showed progression comparing to previous CT. Ultrasonography of the testes revealed variable echogenicity. The tumor markers for germinative tumors (CEA, α-FP, β-HCG) were negative. Biopsy of testes was performed. Urinary steroid profiling was performed to confirm the salt wasting form of CAH. In the treatment 0,5mg of dexametasone was introduced.

Laboratory results

	Laboratory results
Morphology	Ht = 57,4%, Hb= 19,0g/dl
Testosteron	> 15 ng/ml; N: 1,56 - 8,77 ng/ml
FSH	0,16mIU/mL; N: 1,37-13,5mIU/mL
LH	0,16mIU/mL; N: 1,37-13,5mIU/mL
ACTH	> 1250pg/ml; N : 7,2 – 63,3 pg/ml;
17-OH progesteron	>19,2ng/ml; N: 0,2-1,3 ng/ml;
Na	138 mmol/l;
K	4,85 mmol/l;
Cholesterol	231mg/d; N < 190 mg/dl;
Triglycerides	286 mg/dl; N< 150 mg/dl

	Treatment	Results
At discharge from hospital	0,5 mg dexamethasonum	ACTH >1250 pg/ml; Testosteron > 15 ng/ml ; FSH: 0,16mIU/mL; LH : 0,03 mIU/mL;
After 3 months	0,5 mg dexamethasonum	ACTH = 220pg/ml; Testosteron = 11.41ng/ml; FSH 1,33 LH = 0,2 nIu/ml;

Conclusions

This case illustrates that congenital adrenal hyperplasia due to 21-hydroxylase deficiency can progress to chronic complications. Children with CAH require regular screening for complications and metabolic sequelae. It is desirable for all men with CAH to have a testicular ultrasound and routine semen analysis. Patients and their relatives should be well educated and informed about disease.