Late onset 11 Beta Hydroxylase Deficiency: Two cases

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Introduction

Differential diagnosis should include congenital adrenal hyperplasia (CAH) in premature adrenarche patients. Clinically, it is possible to diagnose late onset and simple virilisation CAH caused by 21 hydroxylase deficiency because the criteria are better defined and genetic tests are widely available. But especially late onset 11 Beta hydroxylase deficiency can be very difficult to diagnose because the diagbostic criteria are not well defined and genetic tests are not available for this rare type of CAH. Hereby we present two patients with premature adrenarche who were diagnosed with late onset 11βHE

Cases

	Case 1	Case 2
Age	7 years 9 month	6,5 years
Gender	Female	Female
Compliants/ duration	Axillary hair, adult type sweat- 2 months	Axillary and pubic hair 1 month
Natal	Term, 3250 gr	Term, 3500 gr

physical examination;

	Case 1	Case 2
Height -cm (SDS)	128,3 (0,14)	121,6 (0,53)
BMI (kg/m^2) (SDS)	15,3 (-0,36)	17,2 (0,84)
Midparental height (SDS)	163,5 cm (0,05 SDS)	160 cm (-0,05 SDS)
Telarş	Tanner 1	Tanner 1
Pubic hair	**Tanner 1**	**Tanner 2**
Axillary hair	**(+)**	**(+)**
External genitalya	cliteromegaly Ø, çift açıklık(+)	cliteromegaly Ø, çift açıklık(+)
Other system	Normal	Normal
TA (mm/Hg)	105/60	100/60

Laboratory;

	Case 1	Case 2
LH (µIU/l)	<0,07	<0,07
FSH (µIU/l)	2,55	1,3
E2 (pg/ml)	13,3	<12
DHEAS (0-45 µg/dl)	**121,9**	**159,2**
T. Testosteron (0-20 ng/dl)	**25**	**52**
17OH-P (0-1,5 ng/ml)	0,086	0,21
Bone Age (PH)	8 y 10 mo**(157,9 cm)**	8 y 10 mo **(155,1 cm)**

ACTH stimulation test;

	Case 1	Case 2
ACTH pg/ml (0-46)	15,2	34
Cortisol µg/dl (0')	17,6	14,9
Cortisol µg/dl (30')	31,1	36,2
CortiSol µg/dl (60')	37,6	39,5
17OH-P ng/ml (0')	0,086	0,21
17OH-P ng/ml (30')	0,023	0,79
17OH-P ng/ml (60')	0,3	0,96
11-Deoksicortisol ng/dl (0') (0-344)	**22,6**	**40,1**
11-Deoksicortisol ng/dl (30')	237	755
11-Deoksicortisol ng/dl (60')	**499**	**806**

Mutation (CYP11β1)
(Case 1 and Case 2)

Exon 1-R34Q
Exon 7-A386V
Compound heterozygote

24 hr blood pressure monitorisation		
Total systolic hypertension (%)	**19**	**9**
Total diastolic hypertension (%)	**7**	**2**
Day time systolic hypertension (%)	**5**	**3**
Day time diastolic hypertension (%)	**0**	**0**
Nocturnal systolic hypertension (%)	**41**	**17**
Nocturnal diastolic hypertension(%)	**19**	**3**

Treatment; Hydrocortisone (7,5 mg/m2/day)

Discussion and result

Prevalance of late onset 11βHE among premature adrenarche patients is not known in our country. Ten to twenty times increases in stimulation tests should be carefully evaluated and diagnostic work up should include genetic confirmation when necessary. Diagnostic criteria will be reached by this way.

References

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