Assessment of Sexual Identity in Patients with Congenital Adrenal Hyperplasia

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Background

Sex assignment in infancy for patients with disorder of sex development (DSD) is a challenging problem. Some of the patients with congenital adrenal hyperplasia (CAH) have DSD that may affect their sexual identity.

Objective and Method

The goal of study was to assess gender identity in patients with CAH.

In this study, 51 patients with CAH among 240 cases were selected randomly [**21 children (5-14 yr) and 30 adolescents and adults (15 – 37 yr) using two different sexual identity questionnaires for children and adults based on the criteria of Diagnostic and Statistical Manual of Mental Disorders, 5th edition** **The reliability of the questionnaires was determined acceptable by Cronbach's alpha (α) score of 0.955 for adults and 0.726 for children**.

Results

The study patients were 51cases (17, XY; 34, XX), **Age of patients:** 19±10 yr (range, 5 – 37); **XY group:** 20.5±8 (10 – 37) yr, **XX group:** 18.8±11 (5–37) yr **Children group:** 21 cases (15 XX, 6 XY), mean age 9.1±3.1 yr (5 to 14 years). **Adult group:** 30 patients (19 XX and 11 XY) with the mean age of 26.6±6.3 years (17 to 37). **Karyotype and Prader stages:** in table 1; **type of CAH:** in table 2

In all of the children group, compatibility was seen between sexual identity and rearing gender.

In adult group, there were three cases of mismatching between sexual identity and gender assignment composed two females and one male with 21-hydroxylase deficiency (21-OHD) and three girls with 11 hydroxylase deficiency (11-OHD) were assigned as boy.

A 28-year old boy has simple virilizing type of 21-hydroxylase deficiency with karyotype of 46,XY and with complete male phenotype. He tended to play the role of female but changed his behavior when understood the sin of this work and by psychological consultation.

Two girls with salt wasting type of 21-OHD tended to male sexual identity during puberty.

One of them with Prader 3 virilization at birth and genitoplasty at one year of age, had poor compliance for taking her medicines and was neglected by the parents. She had 2 years and 3 years lost of follow-up in different periods and when she came to the clinic, clitoromegaly was noticed and 17-hydroxy progesterone was high,. She aggressively requested to changing her gender. Her behavior changed after 3 month good control of her endocrine status. She undertook plastic nose surgery and accepted to put on girl's clothes thereafter.

Another one experienced a love failure with a boy and has attributed to girls sexual relationship. She also changed her desire with psychological consultation.

One girl with 11-hydroxylase deficiency (11-OHD) (Prader 4)were reared as a boy without diagnosis until she came to our clinic. Her parent and she did not accept to change the gender when she was 6 years of age with a virilized genitalia and 11 year bone age. Now she is 36 years old with short stature. Testosterone injections are needed. His compliance is very poor and she is depressed and unsatisfied with her gender.

Another one with 11-OHD and Prader 5 virilization had good control of hormones. She was reared as a boy due to rejection of surgeons for her operation at infancy. She accepted to change her gender at 12 years of age when breast developed and one menstruation occurred. She is 34 yrs. old now, is married, have two children and is satisfied with her gender.

Two patients with lipoid adrenal hyperplasia who are 13 and 19 years of age have 46,XY karyotype. They have complete female phenotype, rearing gender is female and testes were removed. Hormone replacement therapy has begun for their pubertal signs and they have female sexual identity without any problem till now.

Table 2, Frequency of different types of CAH in study subjects according to age groups and karyotype

Type	5-14 yr			≥15 yr			total
	XY	XX	total	XY	XX	total	
21(OH)	4	10	14	6	14	20	34 (66.7)
11(OH)	1	2	3	4	3	7	10 (19.6)
3B(OH)		2	2		2		4 (7.8)
Lipoid	1	1	2	1		3	3 (5.9)
Total	6	15	21	11	19	30	51

Table 1, Prader stage of virilization in females according to age groups

Age groups	Prader stage, Frequency (percent)						Total
	0*	1	2	3	4	5	
5 - 14	3 (18.75)	3 (18.75)	4 (25)	3 (18.75)	2 (12.5)	1 (6.3)	16 (100)
≥ 15	3 (15)	4 (20)	3 (15)	4 (20)	3 (15)	3 (15)	20 (100)

*, Two XY patients with lipoid adrenal hyperplasia are included

Conclusion

In patients with CAH, gender identity disorder is a rare finding. Good control of endocrine status not to increase the adrenal hormones prevents changing of their gender identity. XY karyotype has no effect on gender identity in XY lipoid adrenal hyperplasia.

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