Masculinizing operation for a female patient with congenital adrenocortical hyperplasia due to 21-hydroxylase deficiency

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MASCULINIZING OPERATION FOR
A FEMALE PATIENT WITH CONGENITAL
ADRENOCORTICAL HYPERPLASIA DUE
TO 21-HYDROXYLASE DEFICIENCY

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A 25-year-old female with simple virilizing type of congenital adrenocortical hyperplasia
due to 21-hydroxylase deficiency was treated surgically with masculinizing operations which
consisted of two-stage procedures. The first procedure was chordectomy associated with
excision of both gonads and female internal genitalia. Eleven months later, the second
procedure consisting of urethroplasty and implantation of testicular prosthesis was performed.
The postoperative course was successful in terms of urination and penile erection.

Key words: Congenital adrenocortical hyperplasia, Ambiguous genitalia, Masculinizing operation

INTRODUCTION

Congenital adrenocortical hyperplasia represents the most common cause of
ambiguous genitalia in female infants. A variety of enzymatic defects exist, which
result in excessive secretion of adrenal androgen. In cases of 21-hydroxylase
deficiency, the treatment with glucocorticoid is started from early infancy after
diagnosis is established from elevated 17-ketosteroids and pregnanetriol levels. In
this paper, an unusual case of 21-hydroxylase deficiency treated in adult age with
masculinizing operation is reported.

CASE REPORT

The patient weighed 2,730 g at birth after the second full term pregnancy of
a 23-year-old woman. Pregnancy was uncomplicated and no medications other
than vitamins were taken by the mother. In spite of ambiguous external genitalia
and hyperpigmentation, abnormalities were not examined and treated by a pediatrion in the early period of life. The

patient was reared as a boy. At 25 years of age, the patient was admitted to our
hospital for examination and treatment of her abnormalities, with a height of
144.7 cm and weight of 47.6 kg (Fig. 1 and 2), and was diagnosed to have congenital
adrenocortical hyperplasia due to 21-hydroxylase deficiency from the elevated
urine levels of both 17-ketosteroids and pregnanetriol. The karyotype was 46 XX.
Because of the patients age, psychological and social sex and external physical sex
in addition to personal wishes, bilateral gonadectomy, excision of female internal
genitalia and chordectomy were performed in the first operation. From the
findings obtained during operation, the correlation between female urethra and
vaginal opening was Prader's type III or IV. Eleven months later, the second
operation which consisted of urethroplasty under the Denis-Browne's method
and implantations of siliconized testicular prosthesis into both scrotal pockets was
performed. Postoperative course was uneventful. Urination in standing po-
Fig. 1. Body profile showing small stature and hyperpigmentation

Fig. 2. External genitalia showing perineal hypospadias and bifid scrotum. Glands could not be palpable in both scrotums and inguinal areas

Fig. 3. Retrograde urethro-cystogram showing new urethral orifice opening at the tip of the glans

sition could be done successful and penile erection was noted with no evidence of a ventral curvature (Fig. 3). Postoperative course of hormonal adjunctive therapy with testosterone enanthate was planned.

**COMMENT**

The variable type of masculinization in 21-hydroxylase deficiency may depend on such factors as the timing of the appearance of excessive androgen and endorgan sensitivity to androgen. The degree of masculinization varies from only hypertrophy of clitoris to urethral opening at the tip of the glans. So selection of sex which should be accepted and indication of surgical repair depend on the age of the patient at diagnosis, psychological or social sex and severity of masculinization. Our patient, excision of the gonad and urethroplasty to male type were justified to prevent her social and
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psychological trauma, although the patient would be infertile.

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和文抄録

男性化手術を施行した女子先天性副腎皮質過形成 (21-hydroxylase 欠損) の 1 例

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21-hydroxylase 欠損による先天性副腎皮質過形成をもつ25歳女子（戸籍上は男子）症例に対して2度にわたる男性化手術をおこなった。
1度目は卵巢子宮摘出術と索切除術であり、2度目は11カ月後の尿道形成術と両側前壁丸挿入術である。術後、排尿機能と勃起の状態は良好であった。

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