

Congenital Adrenal Hyperplasia Causing Clitoromegaly

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ABSTRACT

Congenital Adrenal Hyperplasia (CAH) is caused by congenital insufficiency of the enzyme 21 – hydroxylase (21-OHD) in the cortisol synthesis pathway. Because of the virilizing effects of androgens over-production, affected girls develop clitoral hypertrophy. Three patients with CAH are discussed below along with their surgical management and follow-up.

Key words: Congenital Adrenal Hyperplasia (CAH). Clitoroplasty. 21-Hydroxylase deficiency.

INTRODUCTION

Congenital Adrenal Hyperplasia (CAH) consists of a group of disorders of adrenal steroidogenesis. Various forms of CAH are transmitted as autosomal recessive trait. Males and females are equally at risk. The average incidence of classical presentation is 1 in 15000 live births.¹ Although the gender of the embryo is determined genetically at the time of fertilization, the gonads do not acquire male or female morphological characteristics until the seventh week of development.² Females with classical 21-OHD, being exposed to excess androgens prenatally, are born with virilized external genitalia.³

The close association between genotype and phenotype makes it possible to predict clinical outcome in affected subject. There is general agreement that the management should aim at creating a normal female anatomy with a minimum of complications and an improvement of life quality.

Three cases with congenital adrenal hyperplasia and details of surgical management are being described in the following report.

CASE REPORT

Case 1

A 35 years old, divorced nulliparous lady, presented with history of hirsutism and enlarged clitoris since adolescence. She was receiving tablet dexamethasone (2.5 mg) once daily. Regarding family history, all her siblings were healthy, except her step-niece who was suffering from the same problem. On examination, patient was found to have masculine look. The height

was 130 cm and she weighed 40 kg. She had male pattern hair growth and underdeveloped breasts. Perineal examination revealed enlarged clitoris of about 2-3 cm, which on arousal increased to 4-5 cm in length. Labia majora, minora and urethral orifice were normal. On bimanual examination, length of vagina was adequate with normal size, mobile uterus. Abdominal and pelvic ultrasonography showed small uterus with endometrial thickness of 0.5 cm. Both ovaries were normal. Both adrenal glands were not visualized. Abdominal CT scan showed mild left adrenal hyperplasia. Buccal smear showed positive barr bodies. Rest of the investigations are described in the Table I.

Table I: The investigations profile.

Investigations (Scrum)	Case-1	Case-2	Case-3	Normal range (iµ/L)
FSH	6.0 miu/ml	76 iu/l	-	2.5 – 12
LH	3.93 miu/ml	9.58 iu/l	-	0.5 – 10.5
Testosterone	3.7 ng/ml	1.9 ng/ml	0.82 ng/ml	0.1 – 1.0
Cortisol	10 ug/dl	1.07 ug/dl	3.8 ug/dl	7 – 20
DHAES	188 ug/dl	114. ug/dl	101 ug/dl	35 – 430
17 OH progesterone	48.8 ng/ml	304 ng/dl	268.8 ng/dl	0.19 – 4.69
Prolactin	10 mg/L	12.00 ng/L	-	1.2 – 20
Estradiol	50 Pg/ml	46.42 Pg/ml	-	30 – 120

Case 2

The second patient was a 14 years old unmarried girl born at home. Her mother noticed that her daughter's genitalia did not look normal. She took her to a pediatrician, who diagnosed her as a case of congenital adrenal hyperplasia. She was treated with dexamethasone tablet (2.5 mg) once daily. Since menarche, she had regular menstrual cycles. On examination, her height was 123.2 cm and weight 33 kg. She had normal secondary sexual characteristics. Perineal examination revealed clitoral enlargement of 2.5 cm, which increased to 3.5 cm on arousal. Abdominal and pelvic ultrasonography showed enlarged left adrenal gland, uterus and ovaries were normal. Abdominal CT scan revealed enlarged left adrenal gland. Buccal smear showed positive barr bodies. Other investigations are shown in Table I. Her family

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history revealed that she had 4 younger sisters out of whom, a 9 years old sister had ambiguous genitalia.

Case 3

A 9 years old girl-child was the sister of patient described in case 2 again with history of home delivery. She was similarly, diagnosed and treated as a case of congenital adrenal hyperplasia by the pediatrician. On examination, her height was 123.2 cm and weigh 26kg. Her menstrual cycles had not started as yet and secondary sex characters were not developed. Perineal examination revealed enlarged clitoris of 3 cm, which increased to 4 cm on arousal. Abdominal and pelvic ultrasonography showed enlarged right adrenal gland, normal uterus and ovaries. Abdominal CT scan confirmed the findings of ultrasonography. Buccal smear showed positive barr bodies and Table I describes the hormonal profile status.

All three patients underwent clitoroplasty with preservation of the neurovascular pedicles under general anesthesia. A traction suture of 3/0 nylon was placed in the glans of clitoris. An incision was made on the lateral phallus perpendicular to the axis of the clitoral shaft, and carried through a 270 degree semicircular arc to the base of the glans as described by Papageorgiou *et al.*⁴ Two longitudinal incisions were made lateral to the dorsal neurovascular bundle. Two crura were identified, clamped and the mid-body of the clitoris was resected. The base of the glans was sutured to the divided corpora with 4/0 vicryl, and proximal and distal ends of the corpora were closed with 4/0 vicryl. The skin was closed with 4/0 vicryl sutures as well. Histopathological examinations of the resected specimens showed normal corporal tissue. There were no early or late postoperative complication. Patients were satisfied with the aesthetical results.

DISCUSSION

CAH poses many challenges, especially for women which include the issues of genital surgery, disclosure, informed consent, weight, and general well-being not to mention having a chronic condition that is life-threatening. The salt wasting form is usually recognized in the neonatal period when infact all decisions concerning gender should be made. Non-classical forms are characterized by more subtle somatic manifestations of androgen excess.⁵ Patients presenting with CAH are very rare. The patients may either be born with ambiguous genitalia⁶ or have late onset features similar to the patients under discussion.⁷

The management of patients with genital anomalies is a complex problem. The goal remains to correct the visible anatomical anomalies, creating an appearance corresponding to the gender, and a function enabling the individual to lead a normal life, including sexual function

and, if possible, reproduction. Clitoral reduction, especially in an adult, is a procedure which often leaves the glans clitoral without the capacity for tactile sensation.⁸ Recent investigations and reports on long-term results indicate that vaginal orgasm is more of an exception than the rule, so that for women, preservation of clitoral sensitivity is essential to a satisfying sexual life. The technique of clitoroplasty should be modified according to the size of phallus. Good cosmetic and tactile results may be achieved by means of selective excision of the corpora cavernosa and lateral clitoral excisions with careful preservation of the neurovascular bundle and the glans.⁹ The patient under discussion underwent clitoroplasty with preservation of the neurovascular pedicles.

More research is now being carried out into the psychological long-term outcomes of women with CAH. Depression and stress are often reported, particularly to do with relationship, weight problems associated with steroid replacement.¹⁰ These patients are reared as females, but have depression due to clitoral hypertrophy, imagining themselves neither female nor male. They were operated to prevent these psychological problems, as body image for women is important.

Multidisciplinary one stop patients centered care with endocrinology, gynaecology, and psychology expert teams are now gradually becoming the cornerstone of care for CAH.

REFERENCES

1. Alizai NK, Thomas DF, Lilford RJ, Batchelor AG, Johnson N. Feminizing genito plasty for congenital adrenal hyperplasia: what happens at puberty ? *J Urol* 1999; **161**:1588-91.
2. Murtaza B, Mahmood A, Shah SS, Saeed S, Abbasi MH, Tamimy MS, *et al.* Male pseudohermaphroditism. *J Coll Physicians Surg Pak* 2005; **15**:653-5.
3. Forest MG, Recent advances in the diagnosis and management of congenital adrenal hyperplasia due to 21-hydroxylase deficiency. *Hum Reprod Update* 2004; **10**:469-85.
4. Papageorgiou T, Hearn-Stokes R, Peppas D, Segars JH. Clitoroplasty with preservation of neurovascular pedicles. *Obstet Gynecol* 2000, **96**: 821-3.
5. Melissa L. A support group's perspective. *BMJ* 2005; :330-4.
6. Malik R, Ghafoor T. Congenital adrenal hyperplasia. *Pak Armed Forces Med J* 2004; **54**:277-8.
7. Ali L. Congenital adrenal hyperplasia (late onset) due to 21-hydroxylase deficiency. *Rawal Med J* 2003; **28**: 25-7.
8. Oyama IA, Steinberg AC, Holzberg AS, Maccarone JL. Reduction clitoroplasty: a technique for debulking the enlarged clitoris. *J Pediatr Adolesc Gynecol* 2004; **17**: 393-5.
9. Zaparackaite I, Barauskas V, Nielsen HO, Jokela R. Adrenogenital syndrome: feminizing genital reconstruction. *Medicina* 2002; **38**: 706-11.
10. Morgan JF, Murphy H, Lacey JH, Conway GS. Long term psychological outcomes of women with congenital adrenal hyperplasia: cross-sectional survey. *BMJ* 2005; **330**: 340-1.

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