CONGENITAL ADRENO-GENITAL SYNDROME

by

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Introduction

Congenital Adreno-genital Syndrome due to 21 hydroxylase deficiency is the most common cause of genital ambiguity in the new born with few cases occurring after puberty. Early diagnosis and correct initial gender assignment continue to be a problem which can be minimised by careful inspection of genitalia at birth and appropriate laboratory studies.

This study presents 5 cases of 21 hydroxylase deficiency, their mode of presentation, clinical and laboratory evaluation and treatment.

Material and Methods

This study comprises of 5 cases—Cases 4 and 5 who were brought for the identification of the correct sex, 2 of the subjects for primary amenorrhoea ,Cases 1 and 2) and 1 (Case 3) who came for sex conversion as the psyche was of a male but was reared as a female. Cases 1 to 3 had findings of virilisation and deepening of voice which became manifest after puberty. All 5 cases had clitoromegaly defined as on stretch greater than 1.5 cms and diameter greater than 1 cm. (Figs. 1, 2 and 3 of Case 2).

Table I depicts the presentation and clinical observations of all 5 cases.

Laboratory investigations include buccal smear for sex chromation, maturation index from vaginal cytology and karyotyping for cases 1 to 3 (Table II) Fig. 4 shows karyotyping of case 2.

Laboratory investigations included for the correct diagnosis are the 17-Ketosteroid values of a 24 hour urine collection before and after dexamethasone therapy. The dose of Dexamethasone ranged from 0.5 to 0.75 mgm tid (Table III).

Laboratory evaluation for cases 4 and 5 could not be done.

Results

All 5 patients had clitoromegaly while adult patients had primary amenorrhoea, deepening of voice, signs of hirsutism and poor development of breasts. Clinical examination revealed the presence of uterus and cervix. Vaginal examination was not done for cases 4 and 5.

Buccal smear and karyotyping of cases 1 to 3 showed presence of X chromatin and normal chromosomal components respectively.

17-Ketosteroid values ranged from 21.1 to 30 mgm and 4.4 to 6.5 mgms before and after dexamethasone therapy respectively.

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TABLE I
Clinical Observations of the 5 Cases

S. No.	Age	Prim. Amenorrhoea	Hirsutism	Breasts	Pubic hair	Clitoromegaly	Deepening of voice	P.V. findings
Case-1	17 Yrs.	+	+	Developed but regressing since 2 Yrs.	++	+	+	Cx. + Ut. + Gonads not felt
Case-2	16 Yrs.	+	+	Not developed	+	+	+	Cx. + Ut. + Gonads not felt
Case-3	20 Yrs.	+	+	Slightly developed	+	++	+	Cx. + Ut. + Gonads not felt
Case-4	1 Yr.	—	—	—	—	+	—	—
Case-5	New born	—	—	—	—	+	—	—

TABLE II
Cytological Tests

Case No.	Barr body	M.I. Vaginal smear	Karyotyping
1.	Positive 32%	70/30/0	46 xx
2.	Positive 30%	80/20/0	46 xx
3.	Positive 28%	50/30/20	46 xx

TABLE III
17-Ketosteroid Test

Case No.	17-Ketosteroids in 24 hr. sample before dexamethasone	17-Ketosteroids in 24 hrs. after dexamethasone suppression
1.	30 mgms/24 hrs.	6.5 mgms/24 hrs.
2.	21.1 mgms/24 hrs.	4.4 mgms/24 hrs.
3.	28 mgms/24 hrs.	5.5 mgms/24 hrs.

Discussion

Congenital adrenal hyperplasia due to 21-hydroxylase deficiency is the most common cause of genital ambiguity in the female.

Adrenal virilism may be congenital and associated with electrolyte disturbance and increased levels of 17-ketosteroid which may be fatal in the first few months of life or later virilism may occur during puberty.

In our study, the first three cases were reared as females and manifested with primary amenorrhoea, hirsutism and clitoromegaly. Cases 4 and 5 had clitoromegaly and were identified at birth as males.

These cases present special problems since children are assigned to a totally inappropriate sex because of errors in diagnosis. Such errors may lead to serious psychological problems. Early recognition of the presence of female pseudohermaphroditism prevents errors in sex orientation. In a study by Roddick and Hammond (1975), 33 patients of congenital adrenal hyperplasia were correct-

ly diagnosed on the basis of elevated 17-Ketosteroids, clinical response to cortisone replacement, response to the Dexamethasone suppression test, buccal smears and karyotyping. In 6 patients, gender assignment was changed after evaluation and therapy.

In this study of 5 cases, the above tests were done to confirm the clinical diagnosis. One of the patients (case 3) underwent sex change. Steroid therapy given to all 5 cases helped to prevent or arrest virilisation and achieve normal growth and development.

Conclusion

A diagnosis of adrenal hyperplasia in patients with ambiguous genitalia is imperative soon after birth since change in the sex of rearing is feasible until 1½-2 years of age after which serious psychiatric consequences can occur. Some of the tests, mainly laboratory, will help establish the diagnosis.

References

1. Roddick, D. H. and Hammond, C. B.: Obstet. Gynec. 45: 15, 1975.

See Figs. on Art Paper VIII