Original article:

Early diagnosis of Intersex disorders in Indian population

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Abstract:

Introduction: The initial goal of the treatment is to determine the appropriate sex of rearing as quickly as possible. The object should not be to reveal the "true sex" of the child but to make a rational decision, based on data containing biochemical and chromosomal studies, steroid metabolism, internal and external genitalia and gonadal histology. So that the sex of rearing, that is commensurate with the most satisfying eventual functional result be assigned to the child.

Methodology: The thesis is study of 30 cases of ambiguous genitalia for a period from January 2001 of June 2003 and their surgical management. The Karyotype can be obtained from a small sample of venous blood or, in the case of a premature baby, from the bone marrow

Results: Prenatal diagnosis of CAH can be achieved on chorionic villous biopsy (CVB) taken at 09-10 weeks of gestation. The laboratory testing involves, determination of the genetic sex and if the foetus is female, DNA test has to be done to establish whether the baby has CAH or not.

Conclusion: Prenatal diagnosis of CAH can be achieved on chorionic villous biopsy (CVB) taken at 09-10 weeks of gestation.

Introduction:

The initial goal of the treatment is to determine the appropriate sex of rearing as quickly as possible. The object should not be to reveal the "true sex" of the child but to make a rational decision, based on data containing biochemical and chromosomal studies, steroid metabolism, internal and external genitalia and gonadal histology. So that the sex of rearing, that is commensurate with the most satisfying eventual functional result be assigned to the child.[1]

Any delay or an incorrect decision may result in death in early infancy from an uncorrected metabolic disorder, the appearance of inappropriate secondary sexual characters at puberty, malignant degeneration of gonads or severe emotional difficulties for child.[2]

With this knowledge in the background, once basic data and investigations are available, it is possible to assign a proper sex to the child by reconstructive surgical procedure.

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Methodology:

The thesis is study of 30 cases of ambiguous genitalia for a period from January 2001 of June 2003 and their surgical management. The Karyotype can be obtained from a small sample of venous blood or, in the case of a premature baby, from the bone marrow. Lymphocytes are usually incubated for 24 hours, at which time the .culture is inoculated with colchilcine to arrest mitosis in metaphase. The chromosomes are then seen to best advantage and photographed. A minimum of forty lymphocytes should be examined to be reasonably confident of identifying mosaicism.

A 46XX Karyotype is found in female pseudohermaphroditism, many true hermaphrodites and XX form of gonadal dysgenesis. A 46 XY Karyotype is present in all male pseudohermaphrodites, in the XY form of gonadal dysgenesis and intsome true hermaphrodites. Mosaicism with 46XY and 46XO lines is the rule in mixed gonadal dysgenesis, whereas 46XY /46XX lines or other combinations may be found in true hermaphrodites.

Results:

Table No: 1 GONADAL DISTRIBUTION AND OPERATIVE PROCEDURE IN TRUE HERMAPHRODITES:

Pt. No.	Operative findings			Operative procedure	Sex of rearing
	Uterus	Gonad			
		Left	Right		
1	Normal	Testis	Ovoestis	B/L gonadectomy Clitorovaginoplasty	Female
2	Normal	Ovoestis	Ovoestis	B/L gonadectomy Clitorovaginoplasty	Female
3	Normal	Ovoestis	Ovoestis	B/L gonadectomy Clitorovaginoplasty	Female
4	Hypoplastic	Testis	Ovoestis	Right gonadectomy, HPS correction	Male
5	Hypoplastic	Testis	Ovary	Right gonadectomy, Left orchiopexy, HPS correction	Male
6	Normal	Ovoestis	Ovary	Left gonadectomy, Vaginoplasty	Female

HPS – Hypospadias

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Table No 2 There were two patients of mixed gonadal dysgenesis

- MIXED GONADAL DYSGENESIS:

Patient no.	Age at evaluation	Presenting symptom	Karotype
1	6 years	B/L undescended	46 XY
2	8 months	Ambiguous genitalia	46 X

Table No: 3 TREATMENT IN MIXED GONADAL DYSGENESISI

Patient no.	Gonadal biopsy Left	Right	Operative procedure	Sex of rearing
1	Dysgenetic testis	Testis	B/L gonadectomy+ clitoroplasty	Female
2	Streak gonad	Ovary	Left gonadectomy+ clitoroplasty	Female

Discussion:

There is no WHY of screening all pregnancies to detect CAH in the foetus. Prenatal treatment is only possible for a couple that has CAH in one of their children.

Prenatal diagnosis of CAH can be achieved on chorionic villous biopsy (CVB) taken at 09-10 weeks of gestation. The laboratory testing involves, determination of the genetic sex and if the foetus is female, DNA test has to be done to establish whether the baby has CAH or not.[3]

Once diagnosis of CAH has been established, it is too late to start treatment and virilization will have already taken place. Therefore, treatment must be started as soon as pregnancy has been confirmed and before the diagnosis of CAH in foetus has been established. The dexamethasone given is 20ugm/kg/day divided into three doses in the day. The main purpose of prenatal diagnosis is to indicate the duration of dexamethasone treatment. [4]Dexamethasone is stopped as soon as it has been shown that the foetus is a male (whether affected or not) or an unaffected female.

If a prenatal diagnosis of CAH can be made before 10 weeks, termination of pregnancy is an option that should be offered to the parents. [5]An alternative way of making the prenatal diagnosis is to collect amniotic fluid at 15 to 16 week gestation. The cells are used for chromosomal sexing of the foetus or HLA studies while the fluid can be assayed for 17 hydroxy progeStemne.[43] Aminocentesis causes unintended abortion less frequently than CVS biopsy does.

Conclusion:

Prenatal diagnosis of CAH can be achieved on chorionic villous biopsy (CVB) taken at 09-10 weeks of gestation.

References:

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