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Sex Chromatin Study in Children with Disorders of Sexual Development

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Abstract
In the children with Disorders of sexual development it is necessary to determine the sex of the baby as early as possible. Sex chromatin study was carried out in 60 subjects reared as females; out of that 16(26.67%) were chromatin negative and 44(73.33%) were chromatin positive. Parental consanguinity showed statistically significant association with abnormal sex chromatin. As sex chromatin study is less time consuming it helps to initiate the genetic conselling in DSD subjects.
Keywords: *DSD, sex chromatin, Barr body, consanguinity, CAH.*

Introduction

In clinical genetics it is sometimes necessary to determine rapidly the sex of an individual without waiting for a peripheral blood chromosome analysis. The first question asked by parents and family at the birth of a baby is what is the sex of the baby? It becomes very difficult to clinicians to answer this question and distressing to the parents if the genitals of baby are ambiguous.

The term Disorder of sexual development (DSD), is defined as a congenital discrepancy between external genitalia, gonadal and chromosomal sex[1]. There are several conditions resulting in these disorders and a disease such as the salt-wasting form of congenital adrenal hyperplasia is fatal unless treated promptly and adequately[2]. At times it is impossible to identify the sex clinically.

Hence, early identification of the sex is essential, irrespective of the cause of the DSD, to counsel the parents, for birth registration and to start treatment and discuss the long-term implications of life-threatening disorders.

Cytogenetic analysis is essential to confirm the sex of a newborn but it is time consuming whereas for sex chromatin (Barr body) determination results are available within few hours. For Barr bodies to be present there should be 2 X chromosomes in the genotype. Barr body represents one of two X chromosomes of a female cell. This cell remains condensed and is in inactive state throughout inter phase and appears in the form of sex chromatin seen as a black area on the nuclear membrane[3].

Materials and Methods

The present study was conducted at Genetics division, Department of Anatomy, Grant Govt. Medical College, Mumbai.

Study subjects - Total 60 patients of disorder of sexual development; referred to Genetics division were study subjects. Age of presentation of these subjects were 1 to 144 months.

Inclusion criteria – patients reared as females with suspected disorder of sexual development .

Exclusion criteria- DSD patients reared as males were excluded from this study.

Family history of subjects was asked to study the consanguinity and pedigree charting was done. Pedigree was analyzed to see any pattern of inheritance in the concerned family.

Material

Material used for this study was buccal scraping.

Method

Subjects were asked to rinse the mouth thoroughly with water. Buccal scraping was collected with the help of a spatula, and then spreaded on a glass slide. Slides were dried. Usually 3-4 slides were prepared for each patient. Then slides were fixed in alcohol and stained with toludine blue solution. Slides were studied for the presence or absence of Barr body.

Observations and Results

Table no 1: Distribution of study subjects according to age at presentation.

Age group in months	Clinical presentation Suspected DSD (n=60)			
	Freq(n)	%	Mean	S.D
1-36	22	36.67	15.64	54.64
37-72	10	16.67	58.2	49.66
73-108	04	6.66	99	43.81
109-144	24	40	135.5	55.05

Table no 2. Distribution of study subjects according to consanguinity.

	Clinical presentation Suspected DSD (n=60)	
	No	%
Consanguineous	48	80
Non consanguineous	12	20
Total	60	100

Table no 3: Distribution of study subjects according to sex chromatin.

X- Chromatin	Suspected DSD (n=60)	
	Freq.	%
Negative	16	26.67
Positive (Normal sized)	38	63.33
Positive (Double)	02	3.33
Positive (Triple)	01	1.66
Positive (Large)	02	3.33
Positive (Small)	01	1.66

Discussion

In developing countries like India, birth of a baby with DSD is a social stigmata. It is necessary for the parents to know the sex of the baby as early as possible. With advancement in cytogenetics and molecular genetics; sex chromatin study is least preferred method of sex determination. Cytogenetics study using karyotype, FISH and molecular studies though available in our country are time consuming as molecular study must be followed by a karyotype.

Age of presentation of DSD will depend upon the degree of dysfunction caused[4]. In the present study the age of presentation of DSD for genetic testing is from 1 to 144 months with maximum 40% of patients, followed by 36.67% in the age group of 1 to 36 months.

Consanguinity is defined as the marriage between close relatives. Consanguinity may result in the homozygous condition for recessive autosomal/ deleterious genes. The incidence of consanguinity reported in India is 5–60% and uncle-niece and first cousin are the more commonly occurring relationships in Indian population[5][6].

Congenital adrenal hyperplasia is inherited as autosomal recessive disorder is one of the cause of DSD. In the present study 12(20%) of the subjects were non consanguineous and 48(80%) of the subjects were consanguineous.

Out of 60 subjects studied 16(26.67%) were negative for sex chromatin; Out of 44(73%) sex chromatin positive subjects, double Barr body was seen in 2(3.33%), triple (1.66%),large Barr body in 2(3.33%) and small Barr body (1.66%) of subjects.

When the association of abnormal X- chromatin was studied with the presence of consanguinity, the test was statistically significant with 'p' value less than 0.05. Which indicates that the consanguineous subjects are at more risk of DSD than the general population.

Conclusion

Mosaicism is not detected by sex chromatin study; but sex chromatin study provides result within few hours and helps to initiate the genetic counseling of parents regarding child's medical condition, but must be followed by Karyotyping.

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