



## Cytogenetic Study in Patients with Abnormalities of Genital System

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### Abstract

*This study includes 42 referred cases for cytogenetic analysis with observable genital abnormalities. These cases were further divided into suspected hermaphrodite, hypogonadism in males, swelling in inguinal region and so on. These were subjected to karyotyping studies. Out of 42 subjects 34 (80.95%) revealed normal karyotype, 3 (7.14%) cases of numerical, 1 (2.38%) structural, 1 (2.38%) mosaic, 3 (7.14%) case of sex reversal abnormality were observed.*

**Keywords:** Ambiguous genitalia, cytogenetics, Barr body, Klinefelter syndrome.

### Introduction

Cytogenetic is the important investigation in the diagnosis and evaluation of genetic disorders in congenital anomalies. Disorders of Sex Development (DSD) is the congenital abnormality in which a person whose sex chromosomes, genitalia and/or secondary sex characteristics are determined to be neither exclusively male nor female.

Genital anomalies are estimated to occur in 1 in 4,500 births. The most common cause of ambiguous genitalia in the newborn is congenital adrenal hyperplasia (CAH), it occurs with the frequency of 1 per 15,000 live births.

A newborn with ambiguous genitalia needs prompt evaluation to detect life-threatening conditions (e.g., salt-losing crisis in congenital adrenal hyperplasia [CAH]) and gender assignment. Sex assignment in

these children continues to be a challenging diagnostic and therapeutic problem.

### Materials and Methods

Total 42 referred patients, suspected of having sex chromosome abnormality were studied. Age group of patient studied was newborn to 25 years.

History of patient was taken, pedigree charting done. Physical examination of patient was carried out.

2 ml of venous blood was collected in heparinised syringe under all aseptic precautions. Culture was set, harvesting was done at metaphase stage and slides were stained using GTG banding technique. Metaphase spreads were analysed with the trinocular microscope and Karyotypes were reported using software.

**Results**

**Table no. 1** Physical characteristics with X-chromatin study

Case no.	Age group	Local examination	Phenotype	X-chromatin
1 to 8	0 -10 yrs	Clitoris is enlarged. Labia majora and minora were split No any testis (mass) in scrotal region.	Ambiguous - 4	Positive
			Male- 4	Negative
9 to 30	11-16 yrs	Hypopigmentation of scrotum. Axillary and pubic hairs were sparse	Male	Negative -21
				Positive - 1
31 to 36	17 -25 yrs years (Phenotype - male)	Gynecomastia ,Height was abnormally more; 2 standard deviations. History of no night falls. Axillary and pubic hairs absent.	Males	2 Barr bodies were seen in one slide and rest showed single Barr body
37 to 42	17 yrs to 25 years (Phenotype female)	Ambiguous external genitalia . Male voice. Muscles well developed Reared as females	Females	Barr body positive in all 4 cases



**Figure 1** A newborn with ambiguous genitalia

**Table no.2-** Chromosomal complements reported

Age group	Clinical symptoms	Karyotype	Frequency	Percentage
0 to 10 yrs	Ambiguous genitals	46,XY	04	50
		45,XO	01	12.5
		46,X,i(X)(q10)	01	12.5
		46,XX	02	12.5
Adolescents 11 to 16 yrs	Hypogonadism (Phenotype – male)	46,XY	21	95.5
		46,XX	01	4.5
17 yrs to 25 years (Phenotype male)	Suspected Klinefelter syndrome (Phenotype – male)	46,XY	04	66.6
		47,XXY	02	33.3
17 yrs to 25 years (Phenotype female)	Pseudohermaphrodites (reared as females)	46,XY	02	33.3
		46,XY[18]/46,XX[2]	01	16.6
		46,XX	03	50

**Discussion**

We have studied total 42 subjects out of that; 34 (80.95%) revealed normal karyotype, 3 (7.14%)

cases of numerical,1 (2.38%) structural, 1(2.38%) mosaic, 3(7.14%) case of sex reversal abnormality were observed.

In the age group newborn to 10 years, out of 8 subjects, 4 showed normal male karyotype 46,XY and 2 showed normal female karyotype 46,XX. One is Turner syndrome and other is Turner variant. Adolescent patients presented with signs of hypogonadism one out of them was revealed with 46,XX karyotype whereas rest were genotypically males. In the suspected Klinefelter syndrome 4 were normal males and 2 cases reported as having Klinefelter syndrome. 2 cases of adult hermaphrodites were genotypically males and 1 was mosaic. The genitalia were due to excess of testosterone that was either secreted by adrenal gland or by testicular tissue.

**Conclusion**

Early referral is very important in the genetic diseases. Careful examination of external genitalia with emphasis on gonadal detection, cytogenetic investigations are essential before assignment of the sex. It will help the clinicians in further surgical and medical management of the condition and proper genetic counselling.

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