

Female with 46X+ (marker) chromosome with gonadal dysgenesis: a case report

Keywords: congenital anomalies, primary amenorrhea, phytohaemagglutinin

Introduction

The association of congenital anomalies, primary amenorrhea is often present in individuals with chromosome abnormalities. Chromosomal abnormalities are frequent events with global incidence 7.6 million and in India its prevalence is 66 per 1000 births. Among these congenital malformations are responsible for 15% of perinatal mortality in India. Primary amenorrhea associated with these abnormal karyo types are approx 3.3%.

Abbreviations: PA, primary amenorrhea; CA, congenital anomaly

Aims and objectives

The present study aimed to analyze the chromosomal changes in case referred to the Department of Genetics, KIMS, Narketpally.

Methods

The venous blood was collected from peripheral blood vessel, cultured for 72hour using Roswell Park Memorial Institute Medium (RPMI) with phytohaemagglutinin (PHA). Chromosomes were analyzed after G banding followed by karyo typing.¹⁻⁵

Result

The case referred to the department showed marker chromosome.

Case

A 17year's old unmarried female presented in gynecology outpatient department with complaints of not having attained menarche. There was no family history of congenital urogenital disorders. On examination, she was a hirsute female, with no secondary sexual characters. She had clitoromegaly with well developed labia major. Her ultrasound revealed absent uterus and ovaries. Her renal function tests were normal and her hormonal profile revealed normal estrogen, progesterone, luteinizing hormone and follicle-stimulating hormone values. Chromosome analysis of the patient revealed 46X+ marker karyo type. Further molecular analysis is suggested for confirmation of marker chromosome (Figure 1).⁵⁻¹⁰

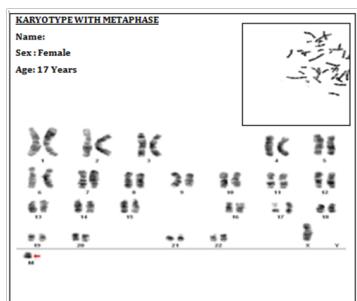


Figure 1 Cytogenetic analysis shows 46 X+ Marker karyo type.

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Conclusion

Chromosomal aberrations represent a major etiologic factor in PA. The role of Genetics in terms of diagnosis risk assessment and genetic counselling is significant and identification of marker chromosomes is important for Genetic counselling. Molecular analysis should be done after cyto genetics for confirmation. After excluding the non genetic causes by clinicians cases should receive prompt referral for genetic study.¹¹⁻¹⁵

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None.

Conflict of interest

The author declares no conflict of interest.

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