ISSN 0972-3757
International Journal of
HUMAN GENETICS
Special Volume

PRINT: ISSN 0972-3757 ONLINE: 2456-6360
Int J Hum Genet, 10(1-3): 71-76 (2010)
DOI: 10.31901/24566330.2010/10.01-3.09

Cytogenetic Analysis of Patients with Primary Amenorrhea

J. Vijayalakshmi, Teena Koshy, Harpreet Kaur, F. Andrea Mary, R. Selvi, V. Deepa Parvathi, R. Bhavani, R. Vikram Jayanth, P. Venkatchalam and Solomon F. D. Paul

Department of Human Genetics, Sri Ramachandra University, Porur, Chennai, Tamil Nadu, India

KEYWORDS Primary Amenorrhea. Turner's Syndrome. Karyotyping. Chromosomal Abnormality

ABSTRACT Amenorrhea is a normal clinical feature in prepubertal, pregnant, and postmenopausal females. It also accounts for 20% of patients with infertility. The physiology of menstruation and reproduction has a strong corre-lation with the expression of the X chromosome. Thus, the role of genetics in terms of diagnosis, risk assessment, and genetic counseling is significant. The genetic contribution to amenorrhea is studied both at the cellular and molecular level aiming at abnormalities in chromosomes and mutations in genes. The present study aimed at performing chromosomal analysis in patients present with primary amenorrhea (n=140) employing GTG banding. The karyotype results revealed 71.2% (n=101) with normal chromosome composition and 27.8% (n=39) showed chromosomal abnormalities. In patients with abnormal chromosome constituents, 74% (n=29) exhibit numerical aberration and 26% (n=10) showed structural abnormalities. The X-chromosome abnormality was observed in 49% of the subject population which is consistent with results of studies conducted in the past. Also, the involvement of Y chromosome and origin of marker chromosome was confirmed by FISH in four patients.