ISSN 0972-3757
International Journal of
HUMAN GENETICS

PRINT: ISSN 0972-3757 ONLINE: 2456-6360
Int J Hum Genet, 14(3,4): 155-159 (2014)
DOI: 10.31901/24566330.2014/14.03-4.06

Cytogenetic Study of Turner Syndrome and Its Variants

Pankaj Gadhia, Parita Balar, Tanvi Gonawala, Nitisha Parekh, Rachna Patel and Salil Vaniawala

Cytogenetics Unit, S. N. Gene Laboratory and Research Centre, President Plaza-A, Surat 395 001, Gujarat, India

KEYWORDS Chromosome Study. G-banding. Karyotype. 45X. Iso(X). Short Stature

ABSTRACT The principle objective of the present study was to investigate postnatal variants of Turner syndrome by cytogenetic study. Total of 1530 cases were referred to the researchers laboratory for cytogenetic analysis (karyotyping), out of which 61 cases of Turner syndrome (TS) diagnosed between March 2005 and January 2014. The most observed karyotype was classic 45,X (49.2 %) followed by iso(X) and iso(X) mosaic each (9.8 %) and least case of number one (1.6 %) was recorded with ring (Xr). Interestingly two cases of Robertsonian translocation t(13;14) were noticed which are considered to be rare. On the basis of clinical features of TS, such as primary or secondary amenorrhea with short stature, the confirmation was done by chromosomal analysis, karyotyping and FISH.