ISSN 0972-3757
International Journal of
HUMAN GENETICS

PRINT: ISSN 0972-3757 ONLINE: 2456-6360
Int J Hum Genet, 12(2): 93-97 (2012)
DOI: 10.31901/24566330.2012/12.02.03

A Data Profile of Phenotypic Features in 181 Turner Syndrome (TS) Females

Vallabhajosyula Ranganath[*] and Sayee Rajangam[**]

[*]Department of Anatomy, Narayana Medical College, Nellore, Andhra Pradesh, India
[**]Department of Anatomy, International Medical School, MSRIT post, New BEL Road, Bangalore 560 054, Karnataka, India

KEYWORDS Turner Syndrome. Variability. Phenotype. Features. Percentage Frequency

ABSTRACT In the present study is reported the data on the observed phenotype in 181 cytogenetically confirmed Turner syndrome females. The features were categorized into 21 groups (skin, low hair line, webbed neck, cardiovascular system, bossing of forehead, highly arched palate, skeletal defects, micrognathia, build, chest, cubitus valgus, digital anomalies, external genitalia, axillary/ pubic hair growth, breast development, far apart position of nipple, ultrasound findings of uterus and ovary). The total number of the 21 features multiplied for the 181 was 3801. TS females manifested only 25% of the features (950/ 3801). Karyotype and its association to the features showed that probands with 45,X (85) manifested 47% (1785/3801) of the features and out of the 950, 45,X showed 50.4% (85) (479/950). The findings confirmed the reported observations that in Turner syndrome, there seemed to be a wide variability in the phenotype.