ISSN 0972-3757
International Journal of
HUMAN GENETICS

PRINT: ISSN 0972-3757 ONLINE: 2456-6360
Int J Hum Genet, 12(4): 319-323 (2012)
DOI: 10.31901/24566330.2012/12.04.04

A Retrospective Study of Balanced Chromosomal Translocations in a Turkish Population

N. Karakus[1], N. Kara[1], S. Tural[1], I. Kocak[2] and M. Elbistan[1]

[1]Department of Medical Biology, Section of Medical Genetics, [2]Department of Gynecology and Obstetrics, Faculty of Medicine, Ondokuz Mayis University, Samsun, Turkey

KEYWORDS Balanced Translocation. Karyotyping. Phenotypic Features. Chromosomal Rearrangements. Cytogenetic Analyses. Recurrent Miscarriage

ABSTRACT The balanced translocations are accepted as chromosomal rearrangements that do not generally reflect any phenotypic evidence. However, phenotypical influences can be seen in children of balanced translocation carriers due to the formation of partial monosomy and partial trisomy of any related chromosome. In this study, 25 cases that detected to have balanced translocation by cytogenetic analyses were evaluated with regard to their phenotypic features. Karyotype analyses of cases were taken out by using conventional peripheral blood culture method. It is estimated that 14 (56%) of these balanced translocation carriers had recurrent miscarriage, 5 (20%) had children with mental retardation, 3 (16%) had infertility, 2 (8%) had amenorrhea and 1 (4%) had mental retardation. When the cases were examined, it is understood that the increase in the frequency of miscarriage is the most frequent phenotypic feature in balanced translocation carriers as a result of the formation of unbalanced gametes.