ISSN 0972-3757
International Journal of
HUMAN GENETICS

PRINT: ISSN 0972-3757 ONLINE: 2456-6360
Int J Hum Genet, 15(4): 199-209 (2015)
DOI: 10.31901/24566330.2015/15.04.05

Congenital Heart Defects and Chromosomal Abnormality

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KEYWORDS Congenital Defects in Heart. Abnormalities in Chromosomes. Genetic Counseling

ABSTRACT Chromosomal abnormality is one of the causal factors in the formation of the congenital heart defects. 65 patients (33 male and 32 female) with heart defects were referred for karyotyping and counseling. Chromosomal abnormalities were detected in 27 (41.5%) and 38 had a normal karyotype. Numerical abnormality was found in 21 (77.8%) and structural in 6 (22.2%), numerical was detected in 14 females and 7 males, and structural in 4 female and 2 male patients. Numerical abnormality was one with 47,XX+13; 2 with 45,X and 18 with 47,XX+21 (11) or 47,XY+21(7). Structural abnormality was derivative 9 in 2, deletion 11q, derivative 14, Robertsonian translocation between 14 and 21 and ring 18 mosaicism in one each. Parental origin of the structural abnormality revealed that two were maternal and one was paternal. In the present study, association could be detected between chromosome 21 and the female probands with chromosomal abnormality and heart defects.