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Analysis of the Chromosomal Deletions

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ABSTRACT Deletion involves loss of part of a chromosome resulting in monosomy for that segment of chromosome. In this paper, a data profile on the detected chromosomal deletions and its association to the phenotype is reported. Data was obtained from 55 probands referred to Division of Human Genetics, from 1974 to 2007. Chromosomal preparations included modified leucocyte microculture method. Deletions were observed in 14 autosomes (2,3,4,5,6,8,9,11,13,15,16,17,18,22) and in X and Y. Deletion in the autosomes was seen in 37 and in X in 13. Deletion, as single cell line was identified in 32 and in mosaic status in 23. Deletion in the long arm of chromosomes was seen in 40 (72.7%) and in short arm in 15 (27.3%) and the break points could be pinpointed in 36. Male to female sex ratio was 1.1:1 (29:26). In 29 males deletion in the autosomes was observed in 24 and in 26 females deletion in autosomes & in X was of equal occurrence (13/26). Chromosomes with deletion seen in both sexes were 3,5,8,9,11,15,18. Deletions were found to be 'de novo' in 9. Deletion was associated to multiple congenital abnormality and or mental retardation (26), amenorrhea (12), bad obstetric history (13) and abnormality in the skeletal (15) and genital systems (15). The analyzed chromosomal deletions and the loss of the chromosomal segments seemed to be associated to a range of clinical conditions and birth defects. The present study, may be for the first time reporting the data on deletion from India.