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Effect of Reciprocal Translocations on Phenotypic Abnormalities

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ABSTRACT The chromosomal disorders make a significant contribution to human mortality and morbidity. Karyotyping allows the identification of various chromosomes involved in a rearrangement. Chromosomal aberrations occur in approximately 1 in 200 live – born infants and the incidence of reciprocal translocations (rcpts) occur as 1/500 live births. Balanced reciprocal translocations can lead to a variety of unbalanced products. In this study, undertaken at the Division of Human Genetics of the Department of Anatomy of St. John's Medical College, Bangalore, 58 cases of reciprocal translocations were collected from the existing data and the results were compiled. The most important observations noted in this study were:

1. The frequency of rcpts was 4.2% among the chromosomal abnormalities identified in the laboratory.
2. The common chromosomes involved in rcpts were chromosomes 1,2,3,5,7,9 and 22. The comparative site of the breakpoints showed preference at 1q, 2p, 5q, 3q, 7p and 9p.
3. Comparison between the distribution of parental carrier status and the cases, which were *de novo*, was explained. Parental carrier status was seen in 18 cases (31.03%) and *de novo* status was seen in the remaining 40 cases (68.97%).
4. Determination of the sex-ratio and the incidence among the affected male/female cases was 1.14:1 showing predictable maternal carrier predominance.
5. A higher clinical correlation between Bad Obstetric History and MR/MCA to various types of rcpts were identified in these individuals.

All these results were correlated and serve as a basis for predictive genetic counseling to these affected individuals and provide clues to the positioning of important genes that may be responsible for human malformations, thus indicating important developmental genes being disrupted during segregation. This study has highlighted for the first time, a profile on rcpts in the Indian population.