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Chromosomal Abnormalities among Children with Congenital Malformations

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ABSTRACT Increase in chromosomal abnormalities is reported in patients referred with birth defects and infertility. This study was aimed to carry out chromosomal analysis for the presence of cytogenetic abnormalities among congenitally malformed children. The karyotypic status could be determined in 176 cases of the 195 malformed children studied. Thirty (about 17%) children exhibited chromosomal anomalies. Among 85 cases with multiple system malformations, 32 (37.6%) showed chromosomal abnormalities and all of them belonged to the category of known syndromes. Chromosomal variants were observed in two children. Down syndrome was the most common syndrome encountered. Evaluation of chromosomal abnormalities is important in understanding the etiology of congenital malformations. Further, a correlation does exist between phenotypic features and the karyotype. Variants are in general, not related directly to the phenotype.