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Numerical Chromosomal Abnormalities in the Malformed Newborns of Goa

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ABSTRACT Several cytogenetic surveys of consecutive births were undertaken at a global level to establish the incidence of aneuploidy and structural chromosomal rearrangements in the human population. A considerable variation in the frequency of chromosomal abnormalities is observed in these studies. The present study was undertaken to examine the incidence of congenital malformations and to record the frequency of numerical chromosomal abnormalities associated with the congenital malformations in Goa. This study revealed an incidence of 19.4/1000 live births. Chromosomal abnormalities were observed in 24.1% of the congenitally malformed newborns, involving 12.7% of numerical abnormalities and 11.4% of structural abnormalities. In most of the cases with numerical abnormalities, the maternal age is advanced (>30 years). It is thus evident that many congenital malformations have genetic etiology. A chromosomal study of each and every child with congenital malformation is recommended in all the pediatric sections of the hospitals for the proper management of such cases.