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Cytogenetic Analysis in Cases with Sex Anomalies

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ABSTRACT The relationship between abnormal chromosomal constitution and sex anomalies has been established since the development of cytogenetic methods. Determination of chromosomal status is the prerequisite for precise diagnosis and counseling of any case with sex anomaly. It is also essential for defining genotypic and phenotypic correlation and understanding the basic mechanism involved in sex determination. In the present study, 156 cases with sex anomalies were investigated. 40 cases (25.6%) showed abnormal karyotypes. These included 10 cases with mosacism, of which 7 were with 45,X/46,XX chromosomal complement; 2 had marker chromosome, 45,X/46,X,+mar; and 1 case showed 46,XY/47,XXY constitution.

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