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Cytogenetic Investigations in 1843 Referral Cases of Disordered Sexual Development From Andhra Pradesh, India

A. Jyothy, K.S.D. Kumar, M. Swarna, M. Raja Sekhar, B. Uma Devi and P.P. Reddy

*Institute of Genetics and Hospital for Genetic Diseases, Osmania University,
Begumpet, Hyderabad, Andhra Pradesh, India*

KEYWORDS Amenorrhea; infertility; Klinefelter's syndrome; sexual ambiguity; sex chromo-somes; Turner's syndrome.

ABSTRACT Chromosomes play an important role in the etiology of disorders associated with sexual development. A referral case study has been undertaken to know the role of chromosomes in the etiology of these disorders and to establish their frequency. A total of 1843 subjects presenting symptoms of abnormal sexual development referred to Institute of Genetics and Hospital for Genetic Diseases, Osmania University, Hyderabad for cytogenetic confirmation and advice over a period of 20 years (January 1979 to January 1999) formed the subjects for the study. Of the 1843 cases suspected for sex chromosome anomalies, 386 (20.95%) showed abnormal karyotypes. Among these abnormalities, Klinefelter karyotypes were found in 2.88% of the cases and Turnerian karyotypes in 4.12% of the cases. The subjects presenting primary amenorrhea and secondary amenorrhoea had abnormal karyotypes in 11.01% and 0.82% of the cases respectively. In individuals presenting sexual ambiguity abnormal karyotypes were identified in 2.12% of the cases. Chromosome abnormalities accounted significantly for the etiology of aberrant sexual development. It is apparent from the study that cases with history of amenorrhoea, infertility and sexual ambiguity need to undergo cytogenetic evaluation as a mandatory routine. Precise diagnosis of these cases has greatly helped for proper management, surgical intervention and counseling the affected.

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