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Chromosomal Abnormalities in 979 Cases of Amenorrhea: A Review

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ABSTRACT Primary amenorrhea refers to absence of spontaneous menarche even after the age of 16 while in secondary amenorrhea, the condition follows a period of normal menstruation. Cytogenetic data in cases with primary (n=852) (PA) or secondary (n=127) amenorrhea (SA) investigated at the Department of Genetics, Dr. A.L. Mudaliar Post Graduate Institute of Basic Medical Sciences, University of Madras, during the 25-year period 1979 to 2004 was reviewed. Routine GTG-band analysis of metaphases from peripheral blood leucocytes revealed the incidence of chromosomal abnormalities in individuals with PA and SA to be 25.82% and 7.09% respectively. In addition to numerical abnormalities, the various structural aberrations of the X chromosome encountered were deletions, isochromosome for the long arm, translocations and ring chromosomes. Ascertainment of the karyotype aided in confirmation of the provisional diagnosis, a better phenotype-genotype correlation to understand clinical heterogeneity and in genetic counseling.