Cytogenetic Findings of Patients with Amenorrhea in Turkish Population: A Retrospective Study

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ABSTRACT We performed a retrospective study, with the purpose of establishing the frequency of chromosomal anomalies in amenorrhea cases referred to our genetic laboratory from hospitals in the Middle Black Sea of Turkey. In this study, 105 cases with amenorrhea were analysed and evaluated by Department of Medical Biology and Medical Genetics. Karyotype analyses of cases were made from their peripheral blood lymphocytes by standard method. Twenty metaphases had been prepared with GTG banding method for each patient was analyzed. When a mosaic karyotype was found in any case approximately 100 cells were examined. Chromosomal anomaly was found in 15 (14.3%) patients. Chromosome anomalies in 15 cases were as follows; 34% 45,X, 13.2% 46,XY (testicular feminization), 6.6% mosaic 45,X/46,XX, 6.6% 45,X,del(Xq21), 6.6% 45,X,del(Xp)(p11.21)/45,X, 6.6% 45,X/46,X,der(X), 6.6% t(X;6), 6.6% t(X;14), 11.76% 46,X,i(X)(q)(10)/45,X. The most common chromosomal abnormalities in cases with amenorrhea were monosomy X and different structural abnormalities of X chromosome respectively.