



# Cytogenetic analysis of 4216 patients referred for suspected chromosomal abnormalities in Southeast Turkey

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**ABSTRACT.** We reviewed cytogenetic studies performed on 4216 patients who were referred to the Cytogenetics Unit at Dicle University Hospital, Diyarbair, Southeast Turkey, between 2000 and 2009. The cases were grouped according to the reason of referral for cytogenetic analysis. The frequencies of the different types of numerical and structural abnormalities were determined, and the relative frequency of cases with abnormal karyotypes was calculated in each group. The most common reason for requesting cytogenetic testing was referral for Down syndrome and for repeated abortions. The highest frequencies of abnormal karyotypes were found among cases that were referred due to suspicion of Down syndrome (84.8%). Among the chromosomal abnormalities, sexual chromosomal abnormalities were found in 239 cases (17.6%), and Klinefelter syndrome was the most frequent sex chromosomal abnormality. Autosomal abnormalities were found in 1119 cases (82.4%), and Down syndrome was the most frequent autosomal chromosomal

abnormality. In conclusion, the high rate of chromosomal abnormalities (32.2%) found in this population demonstrates the importance of cytogenetic evaluation in patients who show clinical abnormalities. This is the first report on cytogenetic testing in the southeast region of Turkey. This type of study provides a basis for determining the risks of recurrence and for deciding on clinical treatment and genetic counseling.

**Key words:** Cytogenetic; Chromosomal abnormalities; Genetic counseling; Turkey