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Cytogenetic Analysis in Down Syndrome

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ABSTRACT Clinically diagnosed Down syndrome cases are referred for karyotyping and counseling. Data on incidence patterns of the 3 cytogenetic types of Down syndrome from the cases seen during the duration of 35 years is presented. Chromosomes were examined after G-banded technique of peripheral lymphocyte cultures. For each patient, in addition to the detailed case history in the proforma 15 metaphases were examined and in cases of mosaicism, the count was increased to 25 to 50 metaphases for analysis. A total of 874 cases were confirmed to have the karyotype of Down syndrome. 509 were male probands (58.24%) and 365 (41.76%) were female cases. The most common was free trisomy 21 in 759 (86.9%), translocation in 77 (8.8%) and mosaicism in 38 (4.3%) cases. Robertsonian translocation 14;21 (48;62.34%) was prevalent among the 77 cases with translocation and the remaining 29 cases had the translocation of chromosome 21 either to chromosomes 1 (1) or 15(2) or 21 (26). The sex ratio has indicated the prevalence of the males for the total sample (1.41:1) (509: 365) as well as for the 3 basic cytogenetic types of DS. Included in the male trisomy 21 are the 2 cases with Klinefelter syndrome (KFS)(48,XXY+21) and one with *de novo* Robertsonian translocation between chromosomes 13 and 14.