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Cytogenetic Evaluation of Down Syndrome: A Review of 1020 Referral Cases

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ABSTRACT A retrospective analysis was performed on 1102 cases with a provisional diagnosis of Down syndrome referred to the Department of Genetics, Dr.ALMPGIBMS, University of Madras during the period from 1979 to 2006. Cytogenetic analyses confirmed the diagnosis in 1020 cases (92.6%). Among them, regular (free) trisomy 21 constituted 83.82 percent. Mosaicism was recorded in 10.78% and Robertsonian translocations in five percent of cases. The translocation was of *de novo* origin in about 50 percent of the individuals where families had been investigated. Trisomy 21 was associated with structural and numerical chromosomal anomalies in one case each. A tandem 21;21 rearrangement and a familial 13;21 Robertsonian translocation with mosaicism for Y chromosome were seen in two other cases. The mean maternal age was higher in regular trisomy 21 (25.08 years) than in translocation (22.83 years) cases. An excess of males was seen in all groups except in the translocation group where the male:female ratio was 0.93. This paper summarizes the chromosomal abnormalities and the clinical features seen in these patients.