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Cytogenetic Findings and Risk Factors for Down Syndrome in Punjab

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ABSTRACT The aim of the present study was to analyse karyotypic pattern, clinical features and factors responsible for the risk of Down syndrome (DS). Chromosomal investigations were done on 114 cases of Down syndrome referred to the Department of Human Genetics, Guru Nanak Dev University, Amritsar. Among 114 cases, 100(87.71%) showed free trisomy 21, mosaicism was present in 6(5.26%) cases and translocations were seen in 3(2.63%) cases. A case of double aneulpoidy was also seen. Average maternal age at the time of birth of DS child was 27.5 years. We found that 25.44% mothers experienced one miscarriage before the birth of a DS child and 7.89% had death/still birth. Average age of DS child referred to the Department was 3.5 years (44 months 6 days) and most of them were either first or second born. In the present study, 49.12% of DS children were diagnosed within first year of life. About 14.9% parents of DS were daily wagers. Birth of DS children was higher in the month of February and least in the month of October. Early diagnosis, karyotyping and awareness about screening tests can prove helpful in decreasing genetic burden.