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Maternal Age and Chromosomal Profile in 160 Down Syndrome Cases – Experience of a Tertiary Genetic Centre from India

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KEY WORDS Down syndrome; maternal age; nondisjunction; translocation; mosaicism

ABSTRACT The present study reports the correlation of maternal age and chromosomal aberration found in 160 Down syndrome cases. The clinical diagnosis of Down Syndrome was evaluated with reference to cytogenetic profiles and maternal age, which revealed that 79% of mothers were below the age of 35 years. The cytogenetic study was conducted using peripheral lymphocytes culture for G-banding and High Resolution Banding of chromosomes. The trisomy 21 due to disjunction effect of chromosomes was predominant as 88.75% of Down syndrome revealed free trisomy 21. Among the advanced (>35 years) age group of mothers, however it was 95.5%. As most pregnant women in our country, while seeking the antenatal or perinatal care are in the younger age group, the routine pregnancy monitoring in healthcare is emphasized as a preventive strategy.

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