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Cytogenetic Profile of Individuals with Mental Retardation

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ABSTRACT The incomplete development of mental capacities and associated behavioural abnormalities are referred to as mental retardation. It is single largest neuropsychiatric disorder in every civilised society affecting 2.5-3.0% of the total population. Chromosomal abnormalities are the important cause of mental retardation. Cytogenetic investigations were carried out on 143 mentally subnormal individuals that were referred to the Centre for Genetic Disorders, Guru Nanak Dev University, Amritsar, India, during 1996 to 2002. These cases were referred mainly as suspected Down syndrome, delayed milestones, mental retardation, etc. The age group of the patients ranged from 1 month to 18 years. Interestingly, maximum number of patients i.e., 58/143 (40.5%) were the firstborns and the average maternal age was 27.6 years. Free trisomy 21 was found to be the most frequent autosomal aberration, both amongst males and females (45.4% males, 18.8% females). It was seen in 92/143 (64.3%) cases while translocations were seen in 2.7% cases. The latter included 45,XY,+t(13;14), 46,XY,+t(14;21), 45,XX,+t(14;21) and 46,XX,+t(14;21) karyotypes.

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