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Chromosomal Abnormalities in Mental Retardation: Indian Experience

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ABSTRACT At a Tertiary Genetic Centre, children with mental retardation (MR) (also referred as intellectual disability) and associated developmental disabilities were investigated for genetic diagnosis which is important in prevention and genetic counseling while offering the risk of recurrence to the family. A prospective and retrospective cytogenetic study was conducted on 1760 MR cases for chromosomal abnormalities using routine GTG and high resolution banding methods of karyotyping. Out of 1760 MR cases, 555 cases showed abnormal chromosomal constitution (31.5%), and males were more than females (2.1: 1). Numerical chromosomal abnormalities were detected in 40.4% (224 of 555), out of which autosomal abnormalities were 36% (199 of 555) and sex chromosomal abnormalities were 4.5% (25 of 555). Structural chromosomal abnormalities were detected in 52% (289 of 555), out of which autosomal abnormalities were 28.6% (159 of 555) and sex chromosomal abnormalities were 29.5% (164 of 555), with some having both numerical-structural (7.6%) and autosomal-sex abnormalities (1.4%). The chromosomal study revealed Down syndrome as the most common chromosomal abnormality i.e. 45% (250 of 555). The children varied from mild to severe mental retardation with and without multiple congenital anomalies and dysmorphism. A few genetic syndromes with characteristic clinical features were also confirmed due to chromosomal aberrations. Genetic counseling was provided to the family members explaining the importance of recurrence risk, the need for prenatal diagnosis in subsequent pregnancies, along with the management of MR children in Indian set-up.