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Chromosomal Anomalies in Referred Cases with Suspected Genetic Disorders: First Report from Jammu and Kashmir

**Parvinder Kumar, Ashma Gupta, Roopali Fotra, Upma, Sunil Raina, Surbhi Sethi,
T.R.Raina and Subash Gupta**

*Institute of Human Genetic; Human Genetic Research cum Counselling Centre, University of
Jammu, /Govt. Medical College Jammu, Jammu, 180006, Jammu and Kashmir, India*

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ABSTRACT In a four year period (2005-2009), a total of 161 individuals of different age group presenting clinical profile like genetically uncertain syndrome, multiple congenital anomalies, short stature, facial dysmorphism, abnormal behaviour, unclassified mental retardation and Down syndrome were referred to the Human Genetic Research cum Counselling centre, Jammu. Chromosome study was carried out in all the referred cases, when the chromosomal abnormalities were detected in 91 (56.52%) individuals. Besides chromosome study, some non-cytogenetic factors like maternal age, male: female ratio, birth order and consanguinity have also been studied to find out the possible association of these factors with chromosomal aberrations in referred patients.