MULTIPLE XY SYNDROME: A CASE STUDY

ASHMA GUPTA[1], PARVINDER KUMAR[2], SUBASH GUPTA[3] & ABHAY SINGH YADAV[4]

[1,2,3]Human Genetic Research cum Counselling Centre, University of Jammu, Jammu, India

[4]Human Cytogenetics Laboratory, Department of Zoology, Kurukshetra University, Kurukshetra, India

ABSTRACT

The present study is an account of the karyotypic analysis carried out in a three and half year old male child born to a young, healthy, non-consanguineous couple. Clinically the child had undescended testes (Cryptorchidism) with empty scrotal sac. It was an uneventful, full term delivery with no family history of congenital anomalies. Besides cryptorchidism, the child had developmental delay as well as mental retardation. Chromosome study was carried out to rule out the chromosomal cause, if any in the child having gross phenotypic anomalies. The karyotype of the child was found to be 49, XXXYY.

KEYWORDS: Cryptorchidism, Developmental Delay, Chromosome Study