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Ring chromosome 21: A Case Report

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ABSTRACT In this article is reported, the phenotype of the male proband, with the ring configuration in chromosome 21. A 3½-year-old male child was referred for karyotyping, with the chief complaint of mental retardation. The manifested features were the delayed mile stones, long face, large and low set ears, slight epicanthic folds, short little finger, clinodactyly, simian crease, flat feet and hypogonadism. The chromosomal analysis was obtained from the peripheral lymphocyte culture. The karyotype of the male proband was 46,XY,r(21)(p11q21). Chromosome 21 was present from region 11 in the short arm to the region 21 in the long arm. Or the regions lost in 21, were distal to the break points in the short arm: 21p11 to 21pter and long arm:21q22 to 21qter. The phenotype and the genotype were correlated. In effect, proband has had partial monosomy for the distal segments of the short and long arm of 21. The loss of the chromatin material from the short arm of 21, one of the acrocentric chromosomes, is not of any significance, since, the genes are involved in the formation of the nucleoli; whereas, the loss of the long arm may have resulted in the clinical picture of dysmorphogenesis and mental retardation. The parents were counseled.