

Chromosomal Studies in Individuals with Infertility

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Introduction: Infertile individuals frequently have chromosomal alteration. **Objective:** To determine the chromosomal constitution of infertile individuals. **Methodology:** A cross-sectional descriptive study was carried out based on the results of the karyotypes in peripheral blood in infertile individuals. The sample consisted of infertile patients of both sexes, from the province of Villa Clara who were studied in the Cytogenetics Laboratory of the Provincial Center of Genetics, between the years 1991 and 2017. We analyzed the chromosome formulas obtained and the age of the subjects at the time of diagnosis. **Results:** 232 individuals were studied, of which 97 were males and 135 females. 27.1% of the karyotypes were positive and in all, the sex chromosomes were involved. We found numerical chromosomal aberrations in thirty-one men (32%) in relation to Klinefelter Syndrome and variants. Thirty-two women (23.8%) had positive karyotype, where structural aberrations of the X chromosome and mosaicism predominated. The diagnosis of six women with karyotype 46, XY (Androgen insensitivity Syndrome) and two 45, X/46,XY (Mixed gonadal dysgenesis) was significant. The average age at diagnosis in men was 31 years and in women 22 years. **Conclusions:** Karyotyping is an essential study in infertility. The detailed clinical examination of this type of patients would facilitate early diagnosis and adequate therapeutics.

Keywords: Infertility, Chromosomal Studies, karyotype