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Mosaic Status of Lymphocytes in Infertile Men with Klinefelter Syndrome

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ABSTRACT Chromosomal aneuploidies such as Klinefelter syndrome (47,XXY) are the most frequent chromosomal aberrations in infertile men. Normally the chromosomal status of patients is detected by karyotyping of up to 20 metaphase spreads, where low-grade mosaicism may be missed. This study was carried out to test whether Klinefelter patients with 47, XXY karyotype with few characteristic features of KFS may harbour normal 46,XY cell line. We analysed 150 well-spread G-banded metaphases in each Klinefelter patient(n-20). RESULTS: Of 20 patients, 4 patients had (5-12%) normal 46,XY cell line. This had not been identified on analyzing 20 metaphase spreads. CONCLUSIONS: These result suggests that 47,XXY patients with presence of XY cell line may have a higher probability of finding germ cells and isolated foci of spermatogenesis in the semeniferous tubules. This is a good diagnostic and prognostic marker for ART. Therefore infertile Klinefelter patients with low-grade mosaicism with 46, XY normal cell line, are good candidates for for ART/ICSI but in such cases if fertilization occurs it should be followed by preimplantation genetic diagnosis (PGD).