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Case Report: Y Chromosome Microdeletion in an Infertile Patient with Mosaic Klinefelter Syndrome

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ABSTRACT Among genetic factors which contribute about 10-15 percent of male infertility, the most common genetic causes of male infertility are Klinefelter's Syndrome (KS) and Y chromosome microdeletions respectively. Most of the KS patients carry 47, XXY karyotype and almost 15 percent of them are mosaic with variable phenotype. These genetic abnormalities characterized by hypogonadism, azoospermia or oligospremia etc. A 41-year-old male presented with primary infertility with small hard testes and upper limit of FSH and LH. Total azoospemia was showed on semen analysis. 47,XXY/46,XY mosaicism was found in the karyotype analysis from the whole blood culture. Molecular investigation revealed a single deletion of AZFa region (M259 STS in DDX3Y locus). This case illustrates a rare deletion of AZFa region and is differ from previously reported in literature.