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A Journey on Y Chromosomal Genes and Male Infertility

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ABSTRACT In the course of evolution Y chromosome has acquired an important role in sex determination owing to the differentiation of the *SRY* gene from its X homologue. Apart from the functionally specialized *SRY* gene, the Y chromosome harbors several genes responsible for normal fertility. Three different spermatogenic loci namely AZFa, AZFb and AZFc located in the long arm of Y chromosome (Yq) has the vital role in regulating normal spermatogenesis. A microdeletion occurring in any of these regions is attributed to spermatogenic failure leading to infertility in men. Genetic cause of male infertility is found to be 10-15% and the outcome is diverse ranging from no germ cells (Sertoli Cell Only syndrome) to hypospermatogenesis. Genes arrayed in the AZFc region have testis specific expression and deletion of the AZFc region is most common among the Y micro-deletions in men with azoospermia condition. Among the candidate genes of the AZFc region the deletion involving *DAZ* is considered to be the frequent cause leading to azoospermia. The mechanism of micro-deletion is found to be the same in case of AZFa and AZFc region. Among these two loci homologous recombination of flanking, identical sequences leads to micro-deletion. But in case of AZFb region the proximal and distal breakpoints does not exhibit sequence homology although interspersed repeated sequences exist in proximity to the break points.