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Y Chromosome and Male Infertility

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ABSTRACT The Y chromosome though representing only 2-3% of the haploid genome harbours about 107 genes and pseudogenes. Many of these are responsible for spermatogenesis and other male-related functions and deletion of any of these can result in infertility. The association of azoospermia with deletions involving long arm of the Y-chromosome, led to the proposition of an azoospermic factor (AZF). Further mapping of the Y-chromosome resulted in the identification of three regions viz., AZFa, AZFb, and AZFc associated with spermatogenic failure. The microdeletions involving the AZF have been extensively reported to cause male infertility. The genes of the "non-recombing region of Y-chromosome" (NRY) or the "male specific region of Y chromosome" (MSY) play an important role in male fertility. Some important genes identified in this region and associated with male infertility are: SRY, TSPY, USP9Y, DBY, UTY, E1F1AY, RBMY, DAZ, CDY1, BPY2, PRY, and TTY2. Their relative contributions to male infertility are discussed.