

Deletion analysis of Azoospermic Factor c (AZFc) region of Y chromosome and its effect on spermatogenic impairment in infertile and normozoospermic in South India

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Introduction: The Azoospermia Factor c (AZFc) region of Y chromosome undergoes de novo mutation and considered as a significant risk factor for spermatogenic impairment which has been well distinguished in infertile and also in fertile individuals with varied effect. However, the impact of these deletions on etiology of male infertility remains poorly understood Indian heterogeneous population. This AZFc sub deletions increase the risk factor of spermatogenic impairment that results in infertile/sub fertile condition.

Objective: To investigate the correlation between the AZFc subdeletions and spermatogenic phenotypes in admixed population of South India.

Methodology: We recruited the fertile normozoospermic (n= 250) and infertile (n=250) individuals with age group ranging 20-45 years. Semen analysis with sperm function test was performed as per the WHO protocol and five AZFc specific sequence tagged site markers were employed to detect deletions in various gene clusters in AZFc regions namely, DAZ (sY254,sY255), gr/gr (sY1291), b1/b3 (sY1291, sY1197) and b2/b3 deletions (sY1191) by means of standard PCR.

Results: In comparison with other AZFc sub deletions we recorded a high occurrence of gr/gr sub deletion (36.3%) in infertile (n=16) and b2/b3 deletion in fertile (n=12) individuals. Interestingly, we have not observed any changes in sperm count, motility, morphology and vitality among the fertile normozoospermic group. In contrast, azoospermic and oligozoospermic subconditions were predominant in infertile individuals with impaired sperm motility, vitality, and volume. Our current investigation is in accordance with the Han Chinese population study in China/East Asians and also among Japanese population where they have recorded the positive association of gr/gr sub deletions with spermatogenic impairment among infertile.

Discussion: There is a significant differences in the frequency of gr/gr deletions between fertile and infertile groups but not for other AZFc subdeletions. Thus, the Y chromosome AZFc gr/gr deletions result in spermatogenic impairment by reducing the sperm quality. Further, screening of single nucleotide variant and loss of specific gene copy in AZFc region and its effect on male infertility using RFLP approach is in progress.

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