

9th Yazd International Congress and Student Award on Reproductive Medicine with 4th Congress of Reproductive Genetics

Key Lectures

K-53

AZF_C deletions Y chromosome and male infertility

Sadighi Gilani MA.

Department of Urology, Tehran University of Medical Sciences, Tehran, Iran.

Email: masadighi@gmail.com

Male infertility is a common condition with heterogeneous causes. Genetic or epigenetic variations or both contribute up to 15%–30% of cases of male infertility. Genes control a variety of physiologic processes, such as spermatogenesis which occurs in a sequential manner with mitotic, meiotic, and postmeiotic differentiation phases and secretion of hormones. The genetic abnormalities involved in male infertility may be chromosomal (numerical and structural aberrations) or monogenic disorders, mitochondrial DNA (mtDNA) mutations, microdeletion of the Y chromosome, multifactorial disorders, imprinting disorders, or endocrine disorders of genetic origin. The Y chromosome is one of the smallest human chromosomes and microdeletions on the long arm of this chromosome (Yq) is one of the most significant causes of male infertility. Y chromosome microdeletions were present in about

5.2% -12.1% of Iranian infertile men with azoospermia and severe oligozoospermia. The azoospermia factor (AZF) region is further subdivided into 3 non-overlapping regions termed as AZFa, AZFb, and AZFc. Deletion of AZFa and AZFb which are less common are associated with non-treatable azoospermia. Deletions of the AZFc region are most common in men with idiopathic oligozoospermia or azoospermia. Cases with AZFc deletions show a progressive deterioration in spermatogenesis and cases develop azoospermia over a period of time. The clinical outcomes of intracytoplasmic sperm injection (ICSI) for oligozoospermic patients with Y chromosome AZF microdeletion are comparable to those of infertile patients with normal Y chromosomes. For azoospermic men with AZFc deletion microdissection, TESE is recommended with a success rate of sperm retrieval about of 36.3%. In conclusion, the pregnancy and delivery in oligozoospermic patients with AZFc deletion are comparable with other studies, but despite of sperm retrieval in azoospermic patients with AZFc deletion, the chance of pregnancy or delivery in these patients is very low. More attention to surgical points for sperm retrieval and more extensive search for sperm finding and refinement of sperm freezing and ICSI procedure is needed for better results.