

Age and partial AZFc deletion do not predict sperm retrieval in non-mosaic Klinefelter patients

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RATIONALE

The azoospermia factor (AZF) region on the Y chromosome long arm is crucial for spermatogenesis and is known for its unique nearly identical direct and indirect repeat palindromes sequence. In addition to Klinefelter syndrome (KS), the deletion of AZF is another cause for male infertility. In regard to the AZFc partial deletion, *gr/gr* deletion is the most common type followed by *b2/b3* and *b1/b3* deletions. In particular, whether *gr/gr* deletion could cause deleterious effect on spermatogenesis is still an ongoing debate. Based on ethnicity and region, Caucasian (OR=3.721), people from Europe (OR=2.465) and south Asia (OR=2.523) with *gr/gr* deletion had higher risk of male infertility in comparison to those without deletions. In a study of Han Chinese KS men in China, the proportion of partial AZFc deletion is higher than than control population: 27(24.3%) of 111 KS men vs 11(11.7%) of the 94 controls.

OBJECTIVES

Our study aimed to analyze predictive factors for sperm retrieval in our KS men and discuss the association for Y chromosome microdeletion with successful sperm retrieval.

MATERIALS & METHODS

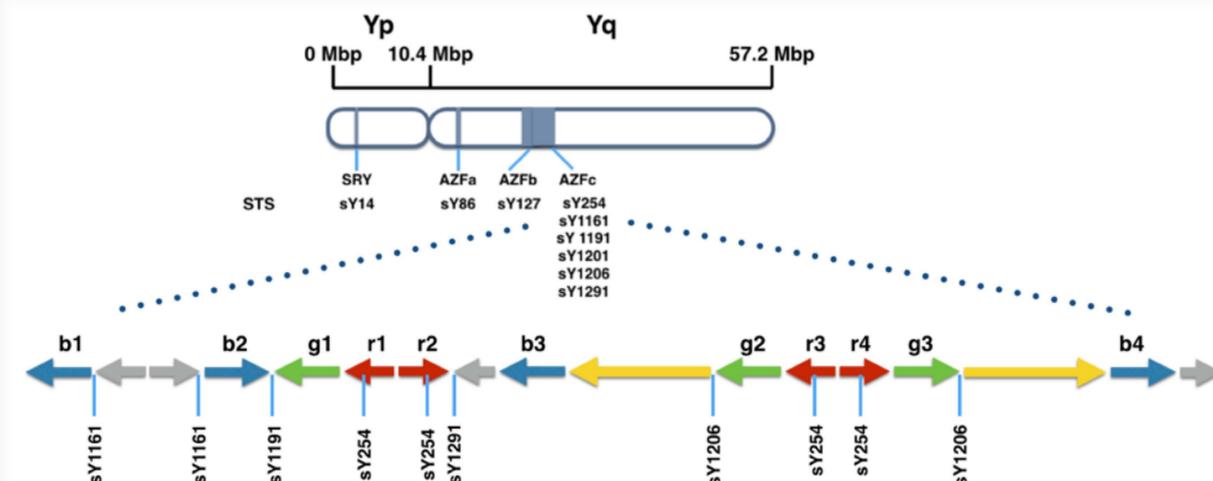
We retrospectively reviewed the period between September 2009 to July 2017, and a total of 53 non mosaic Klinefelter patients receiving microdissection testicular sperm extraction (Micro-TESE) were enrolled in our study. All patients underwent detailed hormone analysis, chromosome karyotyping and Y chromosome microdeletion testing.

RESULTS

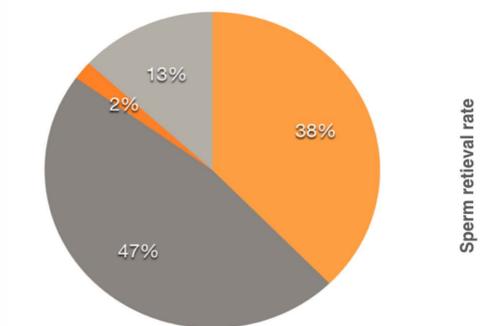
The cohort of 53 azoospermic nonmosaic Klinefelter men with chromosome 47, XXY had a mean age of 36.4 ± 5.4 years (mean \pm SD). Of these 53 men, 8 men (15.1%) had a deletion of one STS marker. Micro-TESE was successful in 20 out of 53 patients, representing 37.7% of cases. Sperm retrieval rate (SRR) for mTESE was 38.1% (8/21) at ages below 35 years and 37.5% (12/32) equal or above 35 years. The partial deletion of azoospermia factor c (AZFc) was noted in 4 (20%) of 20 patients with successful sperm retrieval, including two *b2/b3* and two *gr/gr* deletion cases, whereas 4 (12.1%) of 33 patients with unsuccessful sperm retrieval were noted to have AZFc partial deletion (one *b2/b3*, one sY1206 and two *gr/gr* deletion), but the difference was not statistically significant ($p=0.46$)

CONCLUSION

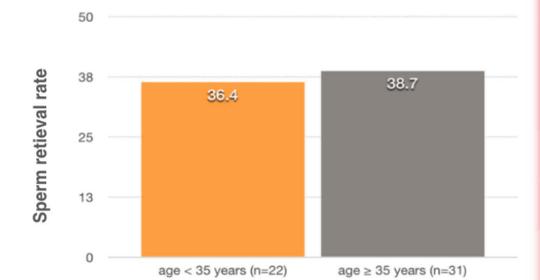
According to present results, age and AZFc partial deletion status should not be the reason to discourage azoospermic nonmosaic 47, XXY men from receiving mTESE.



| | Overall | mTESE (+) | mTESE (-) | p value |
|-----------------------|-----------------|-----------------|-----------------|---------|
| Number | 53 | 20 | 33 | |
| Age (yr) | 36.4 ± 5.4 | 35.9 ± 4.8 | 36.8 ± 5.7 | 0.56 |
| FSH (IU/L) | 34.9 ± 16.8 | 30.8 ± 10.8 | 37.5 ± 19.1 | 0.16 |
| LH (IU/L) | 20.6 ± 12.5 | 17.5 ± 9.4 | 22.5 ± 13.6 | 0.16 |
| Testosterone (ng/ml) | 2.1 ± 1.8 | 2.1 ± 1.8 | 2.2 ± 1.7 | 0.88 |
| Prolactin (ng/ml) | 8.9 ± 3.2 | 9.1 ± 3.5 | 8.8 ± 3.0 | 0.79 |
| Estradiol (pg/ml) | 21.2 ± 10.1 | 19.4 ± 7.2 | 22.3 ± 11.2 | 0.36 |
| AZFc partial deletion | 8 (15.1%) | 4 (20%) | 4 (12.1%) | 0.46 |
| <i>gr/gr</i> deletion | 4 (7.5%) | 2 (10%) | 2 (6.1%) | 0.63 |
| <i>b2/b3</i> deletion | 3 (5.7%) | 2 (10%) | 1 (3.0%) | 0.55 |
| sY1206 del | 1 (1.9%) | 0 (0%) | 1 (3.0%) | 1.00 |



● age < 35 years without partial Y chromosome deletion (n=20)
● age \geq 35 years without partial Y chromosome deletion (n=25)
● age < 35 years with partial Y chromosome deletion (n=1)
● age \geq 35 years with partial Y chromosome deletion (n=7)



● < 35 years (n=22)
● \geq 35 years (n=31)