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Original Research Article

Y chromosome microdeletions in male infertility: prevalence, molecular characteristics and clinical correlates in infertile men

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ABSTRACT

Background: Y chromosome microdeletions within the azoospermia factor (AZF) regions are among the most common genetic causes of male infertility after Klinefelter syndrome. Screening for AZF microdeletions together with karyotyping plays an important role in evaluating men with impaired spermatogenesis before assisted reproductive techniques.
Methods: A total of 242 infertile men attending the andrology and reproductive genetics clinic were evaluated. Clinical assessment, semen analysis, hormonal profiling, karyotyping, and multiplex PCR-based screening of AZFa, AZFb, and AZFc regions using sequence-tagged site markers were performed.
Results: Among 242 infertile men, 80 (33.05%) had azoospermia, 44 (18.18%) severe oligozoospermia, 107 (44.21%) oligoasthenoteratozoospermia, and 8 (3.30%) asthenozoospermia. Y chromosome microdeletions were identified in 32 (13.22%) patients. AZFc deletions were the most common (96.9%), followed by combined AZFb+AZFc deletions and one isolated AZFb deletion. No AZFa deletions were detected. Most affected individuals had azoospermia or oligoasthenoteratozoospermia. One individual with Y chromosome microdeletion also exhibited mosaicism (mos45,X[80]/46,XY[20]). Ten individuals (4.13%) had varicocele with AZFc microdeletions.
Conclusions: Y chromosome microdeletions, particularly AZFc, are highly prevalent among infertile men with azoospermia and OAT. Routine evaluation of karyotype and AZF regions is essential before assisted reproductive techniques (ART) to assess prognosis and prevent transmission of genetic defects.

Keywords: AZFc deletion, Y chromosome microdeletion, Male infertility, Azoospermia, Spermatogenesis

INTRODUCTION

Genetic abnormalities are important contributors to impaired spermatogenesis, and Y chromosome microdeletions represent one of the most frequently identified genetic causes of male infertility.[1] The most frequent complete deletion type is the AZFc deletion (70%–80%) followed by AZFa (0.5%–9%), AZFb (1%–7%), and AZFbc (1%–20%).[2] Genetic testing serves two major purposes in the setting of infertility: determination of heritable conditions that may be passed to offspring and evaluation for conditions which may impact the success of assisted reproductive techniques.[3] Karyotyping and Y-chromosome microdeletion screening are the most common genetic tests used in clinical practice today for male infertility. Currently, Y-microdeletions are the second most common genetic cause of male infertility after Klinefelter syndrome, and genetic testing for AZF deletions has become part of the routine diagnostic work-up of men with azoospermia or severe oligozoospermia.[4,5] Chromosomal anomalies and genetic mutations account for nearly 10–15% of all male infertility cases. Y

chromosome microdeletions have been reported in approximately 11% of azoospermic men and 14% of men with idiopathic severe oligozoospermia, underscoring their significant contribution to male infertility.[6,7] Advancements in assisted reproductive technologies (ART) have enabled men with severe oligozoospermia or azoospermia to father biological children. However, the use of intracytoplasmic sperm injection (ICSI) in men with Y chromosome microdeletions raises concerns regarding the vertical transmission of these genetic defects to male offspring.[8] Genetic factors, including Y chromosome microdeletions, account for approximately 10% of male infertility cases, particularly in men with severe oligospermia or azoospermia. While some studies suggest that the inheritance of AZFc microdeletions may not adversely affect male fertility in sons, the potential for transmission of genetic defects remains a concern. Therefore, it is crucial to conduct thorough evaluations of karyotypes and AZF microdeletions in men with infertility before proceeding with ART, to mitigate the risk of passing on genetic disorders to future generations.

METHODS

This was a descriptive, observational, prospective study conducted in Tata Motors Hospital, Jamshedpur between March 2023 to August 2024.

Study design and setting

This retrospective observational study was conducted in the Genetics Division, Department of Obstetrics and Gynecology, Institute of Kidney Diseases and Research Centre (IKDRC-ITS), Ahmedabad, India, between (insert exact study period, e.g. January 2022 to December 2024).

Study participants

A total of 242 infertile men attending the andrology and reproductive genetics clinic were enrolled after obtaining written informed consent.

Men presenting with infertility and abnormal semen parameters were included. Patients with incomplete clinical records or inadequate samples were excluded.

Inclusion criteria

Infertile men with abnormal semen parameters, including azoospermia, severe oligozoospermia, oligoasthen-oteratozoospermia (OAT), asthenozoospermia, oligoteratozoospermia, or asthenoteratozoospermia, who underwent genetic evaluation, were included in the study.

Exclusion criteria

Patients with incomplete clinical records, inadequate blood samples for molecular analysis, or incomplete genetic investigations were excluded from the study.

Clinical evaluation

All participants underwent detailed clinical examination, semen analysis according to WHO 2021 guidelines, hormonal evaluation (FSH, LH and testosterone), and cytogenetic analysis.

Molecular analysis

Peripheral blood was collected in EDTA tubes. Genomic DNA was extracted by using Qiagen DNA blood mini kit. Multiplex PCR was performed using STS markers covering AZFa, AZFb and AZFc regions together with SRY (sY14) as internal control. A total of 11 AZF-specific STS markers together with the SRY marker (sY14) as an internal control were analyzed for Y chromosome microdeletion screening. Additional AZFc markers (sY153, sY1191, sY1197, and sY1291) were included to improve characterization of partial and extended AZFc deletions. Details of the STS markers, chromosomal locations, primer sequences, annealing temperatures, and PCR product sizes are summarized in Table 1.

AZFa region – sY84, sY86, sY121, AZFb region – sY127, sY134, and AZFc region – sY153, sY254, sY255, sY1191, sY1197, sY1291. PCR amplification was carried out under optimized thermal cycling conditions with annealing temperatures ranging from 56°C to 61°C (Table 1). PCR products were visualized on 2% agarose gels stained with ethidium bromide under UV transillumination. Absence of specific amplification for any STS marker in two independent reactions was considered confirmatory for microdeletion (Figure 1).

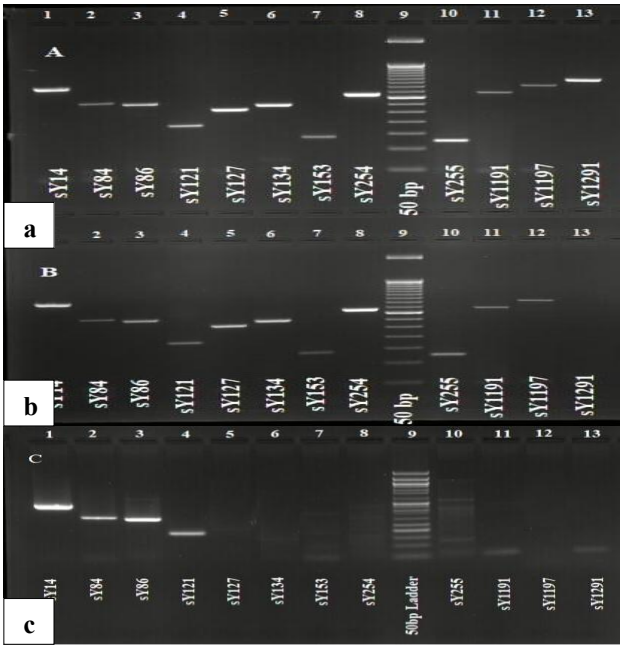

Figure 1: (a) Normal AZF region, (b) AZFc-sY1291 deletion, and (c) AZFb- sY127, sY134 and AZFc - sY153, sY254, sY255, sY1191, sY1197, sY1291 deletion detected.

Statistical analysis

Data were entered into Microsoft Excel (Microsoft Corporation, Redmond, WA, USA) and analyzed using descriptive statistics. Categorical variables were summarized as frequencies and percentages, while continuous variables are presented as ranges. No inferential statistical analyses were performed because the study primarily aimed to describe the prevalence and molecular characteristics of Y chromosome microdeletions among infertile men.

Hormonal assays were performed for FSH, LH, and total testosterone levels using chemiluminescent immunoassay techniques. Chromosomal abnormalities were ruled out by karyotyping.

RESULTS

A total of 242 infertile men were included in the study. The baseline clinical characteristics of the study participants are summarized in Table 2. Among them, 80 (33.05%) had azoospermia, 44 (18.18%) had severe oligozoospermia, 107 (44.21%) had OAT, 8 (3.30%) had asthenozoospermia, 3 (1.24%) had oligoteratozoospermia, and 1 (0.41%) had asthenoteratozoospermia.

Multiplex PCR-based screening for Y chromosome microdeletions across the AZFa, AZFb, and AZFc regions identified 32 positive cases, corresponding to an overall prevalence of 13.22 % among the infertile men studied. The distribution of Y chromosome microdeletions according to semen abnormalities is presented in Table 3. The AZFc region was the most frequently affected and was involved in 31 of 32 cases (96.9%), including 28 isolated AZFc deletions and 3 combined AZFb+AZFc deletions. An isolated AZFb deletion was identified in one patient (3.1%). No AZFa deletions were detected. No AZFa deletions were detected.

Correlation analysis between Y chromosome microdeletion status and semen abnormalities demonstrated that the highest proportion of deletion-positive patients was observed among men with oligoasthenoteratozoospermia (43.75%, 14/32), followed by azoospermia (37.5%, 12/32). Severe oligospermia accounted for 15.625% (5/32) of deletion-positive cases, whereas only one patient with asthenozoospermia (3.125%, 1/32) exhibited a microdeletion. No microdeletions were detected among patients with oligoteratozoospermia or asthenoteratozoospermia (Table 3).

Additionally, ten patients (4.13% of the total cohort) with clinically diagnosed varicocele and normal karyotypes were found to harbor AZFc microdeletions, indicating the coexistence of genetic and anatomical factors contributing to infertility.

Hormonal evaluation demonstrated that FSH levels ranged from 2.68–47.36 mIU/ml, LH from 1.84–42.1 mIU/ml, and testosterone from 113.3 ng/dl to 1173 ng/dl. Elevated FSH and LH were predominantly observed in azoospermic men with extended AZFc or AZFb+AZFc deletions, reflecting impaired spermatogenesis. Testosterone levels were generally within or near the normal reference range; however, a subset of azoospermic men with extensive deletions demonstrated markedly reduced testosterone concentrations (Table 4).

Among the deletion-positive patients, loss of the sY1291 marker was the most frequent molecular finding, either as an isolated deletion or in combination with other AZFc markers. Extended deletions involving multiple AZFc markers (sY153, sY254, sY255, sY1191, and sY1291) were observed in a small number of patients and were generally associated with severe spermatogenic impairment. In contrast, isolated loss of sY1291 was observed predominantly in patients with OAT, severe oligospermia, or azoospermia, indicating considerable phenotypic variability among AZFc deletions. An isolated AZFb deletion involving the sY127 and sY134 markers was identified in one patient, who presented with OAT and a testosterone level of 276.6 ng/dl (Table 4).

Considerable variability in hormonal profiles and STS marker loss patterns was observed among patients with Y chromosome microdeletions. Molecular, cytogenetic, hormonal, and clinical findings were evaluated together for comprehensive assessment of infertility.

Table 1: STS primer, annealing temperature (C) and PCR product size details.

AZF sub regions	STS markers	Chromo-somal location	Gene bank accession no.	Primer sequence (5'-3')	Annealing temperat-ure (C)	PCR product size
SRY (internal control)	sY14	Yp11.3	G38356	F-GAATATTCCCGCTCTCCGGA R-GCTGGTGCTCCATTCTTGAG	56	472 bp
AZFa	sY84	Yp11.221	G12019	F-AGAAGGGTCTGAAAGCAGGT R-GCCTACTACCTGGAGGCTTC	57	327 bp
	sY86	Yp11.221	G49207	F-GTGACACACAGACTATGCTTC R-ACACACAGAGGGACAACCCT	57	320 bp
	sY121	Yp11.222	G38341	F-AGTTCACAGAATGGAGCCTG R-CCTGTGACTCCAGTTTGGTC	61	190 bp

Continued.

AZF sub regions	STS markers	Chromo-somal location	Gene bank accession no.	Primer sequence (5'-3')	Annealing temperat-ure (C)	PCR product size
AZFb	sY127	Yp11.223	G11998	F-GGCTCACAAACGAAAAGAAA R-CTGCAGGCAGTAATAAGGGA	56	274 bp
	sY134	Yp11.223	G12001	F-GTCTGCCTCACCATAAAACG R-ACCCACTGCCAAAACTTTCAA	56	301 bp
AZFc	sY153	Yp11.223	G12004	F-GCATCCTCATTTTATGTCCA R-CAACCCAAAAGCACTGAGTA	60	139 bp
	sY254	Yp11.223	G38349	F-GGGTGTTACCAGAAGGCAAA R-GAACCGTATCTACCAAAGCAGC	60	380 bp
	sY255	Yp11.223	G65827.1	F-GTTACAGGATTCGGCGTGAT R-CTCGTCATGTGCAGCCAC	56	124 bp
	sY1191	Yp11.223	G73809	F-CCAGACGTTCTACCCTTTCG R-GAGCCGAGATCCAGTTACCA	58	385 bp
	sY1197	Yp11.223	G67168	F-TCATTTGTGTCCTTCTCTTGGA R-CTAAGCCAGGAACTTGCCAC	58	453 bp
	sY1291	Yp11.223	G72340	F-TAAAAGGCAGAACTGCCAGG R-GGGAGAAAAGTTCTGCAACG	61	527 bp

Table 2: Baseline clinical characteristics of the study participants (n=242).

Characteristic	Number (%)
Total study participants	242
Azoospermia	80 (33.05)
Severe oligozoospermia	44 (18.18)
Oligoasthenoteratozoospermia (OAT)	107 (44.21)
Asthenozoospermia	8 (3.30)
Oligoteratozoospermia	3 (1.24)
Asthenoteratozoospermia	1 (0.41)
Patients with Y chromosome microdeletions	32 (13.22)
Patients with chromosomal abnormalities	22 (9.09)
Patients with genetic abnormalities (overall)	54 (22.31)
Patients with varicocele and AZFc microdeletion	10 (4.13)

Tables 3: Distribution of Y chromosome microdeletions according to semen abnormalities (n=32).

Semen analysis	Total no. of sample	Y chromosome microdeletion observed	% out of 32
Azoospermia	80	12	37.5
Oligospermia	44	5	15.625
OAT	107	14	43.75
Asthenozoospermia	8	1	3.125
Oligoteratozoospermia	3	0	0
Asthenoteratozoospermia	1	0	0
Total sample	242	32	

Table 4: Clinical and biological parameters of patients with Y chromosome microdeletions (n=32).

Patient no.	AZF deletion pattern	Semen category	FSH (mIU/ml)	LH (mIU/ml)	Testosterone (ng/dl)
1	AZFb+AZFc	Azoospermia	4.33	4.60	537
2	AZFb+AZFc	Azoospermia	4.57	8.42	ND
3	AZFb (sY127, sY134) and AZFc (sY153, sY254, sY255, sY1191, sY1197, sY1291)	Azoospermia	14.21	2.19	1173

Continued.

Patient no.	AZF deletion pattern	Semen category	FSH (mIU/ml)	LH (mIU/ml)	Testosterone (ng/dl)
4	AZFc (sY153, sY254, sY255, sY1191, sY1291)	OAT	28.09	10.30	661.19
5	AZFc (sY1291)	OAT	2.68	6.31	512
6	AZFc (sY1291)	Severe oligospermia	11.58	4.14	452
7	AZFc (sY1291)	Azoospermia	31.47	15.00	317.37
8	AZFc (sY1291)	OAT	13.60	8.21	437.20
9	AZFc (sY1291)	Azoospermia	47.36	14.06	358
10	AZFc (sY1191)	OAT	6.00	8.01	598
11	AZFc (sY1291)	OAT	8.76	7.46	ND
12	AZFc (sY1291)	Azoospermia	7.21	2.26	641.3
13	AZFc (sY1291)	OAT	5.11	2.57	687.2
14	AZFb (sY127, sY134)	OAT	3.90	6.72	276.6
15	AZFc (sY1291)	Azoospermia	10.82	1.84	506.2
16	AZFc (sY1291)	Severe oligospermia	38.57	6.39	512
17	AZFc (sY1291)	OAT	7.35	2.53	492
18	AZFc (sY1191)	OAT	6.25	10.97	>1000
19	AZFc (sY1291)	OAT	11.77	4.59	577
20	AZFc (sY1291)	Severe oligospermia	6.71	6.06	178.5
21	AZFc (sY1291)	Azoospermia	42.20	15.84	288
22	AZFc (sY1291)	OAT	29.67	23.58	199.1
23	AZFc (sY1291)	Asthenozoospermia	4.67	5.53	1839
24	AZFc (sY1291)	OAT	3.49	2.19	244
25	AZFc (sY153, sY254, sY255, sY1191, sY1291)	Azoospermia	21.02	13.33	113.3
26	AZFc (sY1291)	Azoospermia	10.76	10.32	383
27	AZFc (sY1291)	Severe oligospermia	11.06	8.48	299
28	AZFc (sY1291)	Severe oligospermia	13.12	9.87	604
29	AZFc (sY1291)	Azoospermia	6.51	4.79	343
30	AZFc (sY1291)	OAT	11.80	9.07	284
31	AZFc (sY1291)	Azoospermia	13.05	42.10	503
32	AZFc (sY1291)	OAT	7.36	11.45	533

DISCUSSION

Our study confirms that Y chromosome microdeletions represent a significant cause of spermatogenic failure in infertile men, particularly those presenting with azoospermia and oligoasthenoteratozoospermia. The overall prevalence of Yq microdeletions in our cohort was 13.22%, which is consistent with previous reports. For example, Krausz et al and Simoni et al have shown frequencies of 10–15% among azoospermic and severely oligozoospermic men, supporting the robustness of our findings.[9,10] The predominance of AZFc deletions in our series also mirrors the published literature, as several studies have reported AZFc to be the most frequently deleted region, accounting for nearly 70–80% of all microdeletions in infertile men. This consistency strengthens the clinical relevance of our results and affirms the utility of AZFc screening as a primary diagnostic target.

Although no AZFa deletions were identified and only one isolated AZFb deletion was detected in our cohort, previous studies have demonstrated that complete AZFa and AZFb deletions are commonly associated with severe testicular failure and azoospermia.[11-13] Our observation that some men with AZFc deletions had oligozoospermia or OAT, rather than complete azoospermia, is in agreement with these reports and underscores the variable phenotypic expression of AZFc deletions.

However, not all published studies are in complete agreement with our findings. Some reports, particularly from Asian and Middle Eastern cohorts, have suggested lower frequencies of Yq microdeletions, ranging from 5–8% in men with infertility.[14,15] These discrepancies may reflect differences in genetic background, population-specific risk factors, selection criteria for patients, or differences in the number and type of STS markers used for screening. For example, partial deletions within AZFc

(such as gr/gr deletions) are reported at higher frequencies in some populations and are variably associated with impaired spermatogenesis, while in others, they appear to have minimal clinical impact.[16,17] The observation that some sons of fathers with AZFc deletions can have normal fertility also challenges the uniformity of genotype–phenotype correlations and highlights the complexity of Yq deletions.

Our study identified one patient with concurrent chromosomal abnormality and Y chromosome microdeletion, involving mosaicism (45,X/46,XY). While rare, similar cases have been described in the literature, particularly in reports of structural rearrangements of the Y chromosome, ring chromosomes, or mosaic karyotypes in men with infertility.[18,19] Supporting our findings, Colaco and Modi have emphasized that structural anomalies of the Y, particularly involving the Yq11 region, can contribute both to spermatogenic failure and, in some cases, to disorders of sex development.[19,20] The co-occurrence of Yq deletions and karyotypic abnormalities in our patients provides further evidence of the complexity of genetic infertility and the need for combined cytogenetic and molecular analysis.

An additional interesting aspect of our study was the presence of AZFc deletions in men with varicocele. Most prior studies have considered varicocele to be an acquired or surgically correctable cause of male infertility, but the detection of Yq microdeletions in these patients suggests that genetic factors may coexist with anatomical causes of infertility. This finding has been supported by smaller studies reporting Yq microdeletions in men with varicocele, though such cases remain uncommon.[21] On the other hand, some authors have argued that varicocele-associated infertility is primarily mechanical or hemodynamic, with little evidence of underlying genetic contributions. Our results suggest that both mechanisms may coexist in certain individuals, complicating clinical interpretation and management.

The hormonal profiles of our patients also showed wide variation, ranging from elevated FSH in men with severe testicular failure to near-normal values in men with partial deletions or milder phenotypes. This observation is supported by previous reports, which emphasize that while elevated FSH is a common marker of impaired spermatogenesis, it is not uniformly predictive of microdeletions, as residual spermatogenesis may persist in some cases. The variability observed in our cohort is therefore consistent with the heterogeneous endocrine responses reported in the literature.[6,22]

The present study has certain limitations. The analysis was conducted at a single center and included only infertile men referred for genetic evaluation, which may introduce referral bias. Family studies and testicular histopathology were not available for all patients. Furthermore, partial AZFc rearrangements such as gr/gr and b2/b3 deletions were not systematically investigated. Larger multicenter studies incorporating detailed genotype–phenotype correlations are warranted.

Taken together, our findings both support and expand upon existing knowledge. The predominance of AZFc deletions and their variable phenotypic expression is strongly supported by prior studies, whereas the coexistence of Yq deletions with chromosomal abnormalities and varicocele adds further complexity and highlights the importance of comprehensive genetic testing in infertile men. At the same time, the presence of studies with lower reported prevalence or contradictory associations underscores the heterogeneity of Yq deletions across populations, emphasizing the need for population-specific data to guide clinical interpretation.

CONCLUSION

Y chromosome microdeletions were identified in 13.22% of infertile men in our cohort, with AZFc deletions representing the predominant molecular abnormality. Microdeletions were most frequently associated with azoospermia and oligoasthenoteratozoospermia, although substantial phenotypic variability was observed. The coexistence of Y chromosome microdeletions with varicocele and chromosomal abnormalities further emphasizes the complexity of male infertility. Comprehensive genetic evaluation including karyotyping and AZF microdeletion analysis should be incorporated into the routine assessment of infertile men prior to ART, both for prognostic counseling and for reducing the risk of transmitting genetic abnormalities to future generations.

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