Human Reproduction Vol.20, No.8 pp. 2168–2172, 2005 doi:10.1093/humrep/dei034
Advance Access publication April 21, 2005

Cytogenetic, molecular and testicular tissue studies in an infertile 45,X male carrying an unbalanced (Y;22) translocation: Case report

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(Y;autosome) translocations have been reported in association with male infertility. Different mechanisms have been suggested to explain the male infertility, such as deletion of the azoospermic factor (AZF) on the long arm of the Y chromosome, or meiosis impairment. We describe a new case with a *de novo* unbalanced translocation t(Y;22) and discuss the genotype–phenotype correlation. A 36 year old male with azoospermia was found to have a mosaic 45,X/46,X, + mar karyotype. Fluorescence *in situ* hybridization (FISH) showed the presence of a derivative Y chromosome containing the short arm, the centromere and a small proximal part of the long-arm euchromatin of the Y chromosome and the long arm of chromosome 22. The unstable small marker chromosome included the short arm and the centromere of chromosome 22. This unbalanced translocation t(Y;22)(q11.2;q11.1) generated the loss of the long arm of the Y chromosome involving a large part of AZFb, AZFc and Yq heterochromatin regions. Testicular tissue analyses showed sperm in the wet preparation. Our case shows the importance of documenting (Y;autosome) translocations with molecular and testicular tissue analyses.

Key words: AZF deletions/azoospermia/genotype–phenotype correlation/testicular biopsy/(Y;autosome) translocation

Introduction

Azoospermia, when no sperm can be detected on two separate semen samples, is found in up to 10–20% of infertile men (Jarow *et al.*, 1989). Genital tract obstruction and defective spermatogenesis are the principal causes of azoospermia (Jarow *et al.*, 1989). Chromosome aberration is a well-known factor influencing spermatogenesis. Chromosomal abnormalities are present in 7% of infertile men and in 10–15% of azoospermic men (De Braekeleer and Dao, 1991; Van Assche *et al.*, 1996). Among the karyotypic abnormalities found in azoospermic men, sex chromosome abnormalities predominate (mainly 47,XXY genotype). Also, azoospermia may be due to structural abnormalities of the chromosomes. Translocations involving Y chromosome have been reported in association with male infertility (Hsu, 1994). Two possible mechanisms can explain the male infertility. First, the azoospermic factor (AZF), which is critical for spermatogenesis, is located on Yq11 euchromatin and may be affected secondary to a microdeletion, rearrangement or complete loss as a result of the translocation mechanism (Vogt *et al.*, 1996). This mechanism is quite improbable when no deletion loss of AZF loci is observed (Delobel *et al.*, 1998; Pabst *et al.*, 2002) or when the breakpoint on Y chromosome is distal to Yq11 (Delobel *et al.*, 1998; Buonadonna *et al.*, 2002). Thus, a second explanation could be a defective X–Y pairing during meiosis with an abnormal sex vesicle formation and consequently a spermatogenetic arrest (Laurent *et al.*, 1982; Delobel *et al.*, 1998; Buonadonna *et al.*, 2002). We describe here a new case with a *de novo* unbalanced translocation t(Y;22)(q11.2;q11.1) in an infertile man associated with sperm production.

Case report

Clinical report

The patient was referred to our centre for infertility investigations. The patient had absolute azoospermia. No sperm were found in any of the three routine semen analyses. The patient was an apparently healthy 36 year old male. His physical examination was normal, except for a slight testicular hypotrophy (11 and 8 ml). The family history was unremarkable. The elevated serum FSH concentration of 18 IU/l (normal range 3–7) and LH serum concentration of 11 IU/l (normal range 3–8) suggested abnormal

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spermatogenesis. Free testosterone level (280 ng/dl, normal range 450–950) and inhibin B level (24 pg/ml, normal range 80–270) were low.

Conventional cytogenetic analysis

Chromosome analyses were performed on cultured lymphocytes. Standard techniques were used for the preparation of metaphase spreads. The constitutional karyotype was established after R banding.

Fluorescence in situ *hybridization (FISH)*

FISH analyses were performed on lymphocyte metaphase spreads from the patient using standard protocols. The following DNA probes were used for FISH analysis: X chromosome (DXZ1), Y chromosome (DYZ3) and chromosome 14/22 (D14Z1/D22Z1) centromeric specific probes, whole chromosome painting probes for chromosome 22 and Y chromosome and DiGeorge N25 chromosome region probe (Vysis, USA).

Molecular analysis

DNA was extracted from blood lymphocytes. Microdeletion analysis of the Y chromosome was performed using a sequence-tagged site (STS)–PCR approach. Twenty-eight STS corresponding to the three distinct AZF loci were selected. The STS tested were sY84 for AZFa, sY95, sY97, sY169, sY102, sY105, sY109 for the interval between AZFa and AZFb, sY113, sY115, sY117, sY124, sY130, sY134, sY136, sY143, sY142 for AZFb, sY152, sY232, sY156, sY240, sY148, sY249, sY204, sY208, sY254, sY269, sY158 for AZFc, and sY160 for the heterochromatic distal Yq region.

Testicular tissue analyses

Fresh testicular tissue was obtained surgically from the patient. Direct cytological examination and histological examination were performed on testicular tissue.

Immediately after testicular biopsy, the extracted testicular seminiferous tubules were deposited on a sterile glass slide and gently dissected with sterile microscissors in FertiCult medium (Izard *et al.*, 1999). The wet preparation of the suspension was then checked for the presence of sperm under a phase-contrast microscope at ×400 magnification. The morphology of sperm was assessed using Shorr staining. Sperm morphology was evaluated according to modified David's classification (Auger *et al.*, 2001).

For testicular histology, the biopsy specimen was fixed with AFA (75% alcohol, 0.8% formalin and 5% acetic acid). After embedding with paraffin, tissue sections were cut with a thickness of 4 μm. All sections were stained with haematoxylin, eosin and saffron.

Conventional cytogenetic analysis

The cytogenetic analysis was initially interpreted as a 45,X karyotype. Further cytogenetic examination revealed a mosaic 45,X[15]/46,X, + mar[3] with a small chromosomal marker whose origin could not be resolved by conventional analysis (Figure 1A). The father had a normal karyotype.

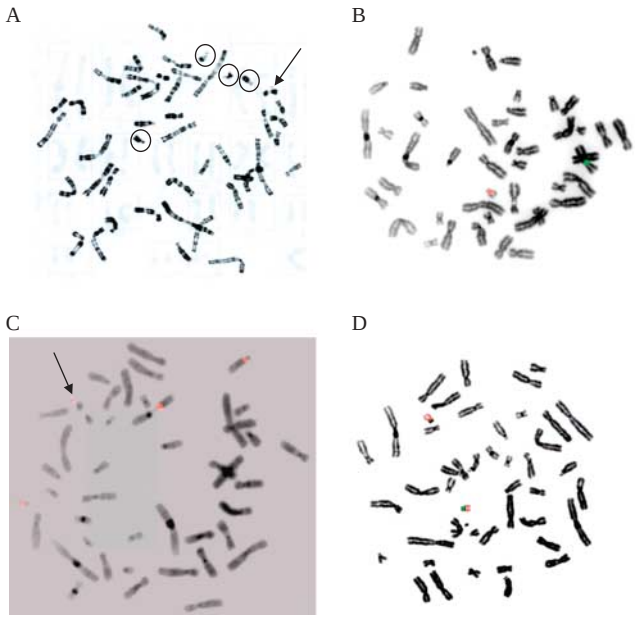

Figure 1. (**A**) Metaphase spread of the propositus after R-banding showing the 46,X, + mar karyotype including four G group chromosomes (circles). The arrow indicates the small marker chromosome. (**B**) FISH analysis with centromeric specific probes for X chromosome (green) and Y chromosome (red). (**C**) FISH analysis with the 14/22 α-satellite centromeric probe showing a signal on the marker chromosome (arrow). (**D**) FISH analysis with a paint specific for Y chromosome (green) and a paint specific for chromosome 22 (red).

FISH analysis on lymphocyte metaphases

Two-colour FISH with the X chromosome centromeric (green) and the Y chromosome centromeric (red) specific probes showed the presence of both centromeres. The Y chromosome centromeric specific probe showed that the hybridization signal was located on a chromosome 22 (Figure 1B).

The 14/22 α-satellite centromeric probe showed signals on both centromeres of chromosomes 14, one signal on the centromere of one chromosome 22 and one signal on the centromere of the small chromosome marker. This suggested that the small marker chromosome included the centromere of one chromosome 22 (Figure 1C).

Two-colour FISH with paint specific probes for Y chromosome (green) and chromosome 22 (red) showed a derivative Y chromosome containing the short arm, the centromere and a small proximal part of the long-arm euchromatin of the Y chromosome and the long arm of a chromosome 22. Thus, the small marker chromosome represented a part of the translocation event corresponding to the short arm and the centromere of chromosome 22 (Figure 1D). On chromosome 22, the breakpoint occurred in the region just below the centromere as confirmed by FISH using the DiGeorge locus probe. This probe hybridized on both chromosome 22 and derivative Y chromosome, indicating therefore that the breakpoint on chromosome 22 was located between centromere and 22q11.2 region (data not shown).

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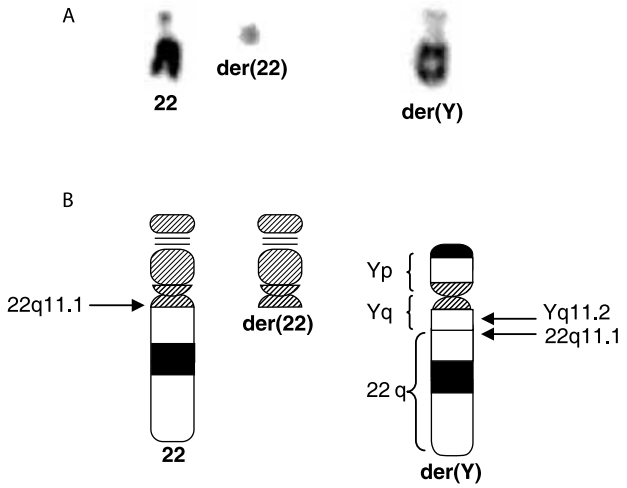

Figure 2. (**A**) Partial R-banding karyotype of the patient showing the translocation t(Y;22). (**B**) Ideogram of G-banding pattern corresponding to the translocation. Arrows show breakpoints.

The results of these hybridization experiments indicated therefore that the patient was a carrier of an unbalanced translocation t(Y;22)(q11.2;q11.1) (Figure 2).

Molecular analysis

STS sY84, sY95, sY97, sY169, sY102, sY105, sY109 gave positive results. The other STS sY113, sY115, sY117, sY124, sY130, sY134, sY136, sY143, sY142, s152, sY232, sY156, sY240, sY148, sY249, sY204, sY208, sY254, sY269, sY158 and sY160 were not amplified. Thus, the breakpoint on Y chromosome was located between sY109 (proximal to AZFb) and sY113 (proximal border of AZFb).

Testicular tissue analyses

A wet preparation of the testicular tissue sample showed the presence of non-motile sperm. Shorr staining showed a normal morphology (the normal morphology rate was 34%) (Figure 3).

The testis histology was analysed in a series of tissue serial sections obtained by testicular biopsies. Histopathological examination showed atrophic and hyalinized tubules with Sertoli cells and few Leydig cells. Neither germ cells nor sperm were observed. Thus the testis histology was interpreted as a Sertoli cell-only syndrome (SCOS).

Discussion

We describe here a new case of an unbalanced *de novo* t(Y;22) translocation in an azoospermic male. This (Y;22) translocation was diagnosed initially as a 45,X conventional karyotype. A male phenotype in the presence of a 45,X chromosome constitution represents a rare and generally unexplained condition. Some of these cases are in fact low-grade 45,X/46,XY mosaics overlooked by routine cytogenetic analysis. Another explanation for a 45,X karyotype is an undiagnosed (Y;autosome) translocation. In our case, a part of the long arm of one chromosome 22 has been translocated onto Y chromosome at band Yq11.1. Because

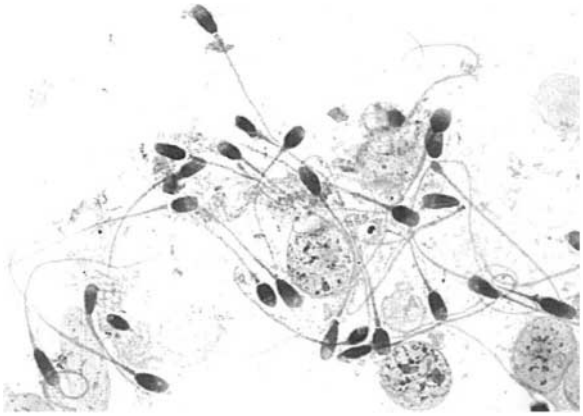

Figure 3. Testicular tissue analysis: wet preparation with Shorr staining showing sperm.

the morphology of the derivative Y chromosome was similar to the morphology of the normal chromosome 22, this translocation was not initially detected. Moreover, the small marker was not easy to detect in conventional analysis due to its small size and was seen more easily with DAPI staining.

(Y;autosome) translocations have been associated with normal and abnormal spermatogenesis. Hsu (1994) reviewed over 130 cases of (Y;autosome) translocations. In the most common form, the heterochromatic region of Yq (Yqh or Yq12) is translocated onto the short arm of an acrocentric chromosome (D or G group chromosome). Other types of (Y;autosome) translocation include balanced and unbalanced reciprocal translocations. A chromosomal breakpoint at band Yq11, as observed in our case, is rarely described. Twenty cases of adult males carrying a (Y;autosome) translocation with a breakpoint at Yq11 region are reviewed in Table I. Since the phenotype may depend on the localization of the Yq breakpoint and on the nature of Yq material lost, it is of great interest to ascertain the Yq breakpoint by DNA molecular studies (AZF screening or interval deletions studies). Deletions of the AZFa, b and c regions are associated with abnormal spermatogenesis from SCOS to hypospermatogenesis (Vogt *et al.*, 1996; Krausz *et al.*, 2003; Simoni *et al.*, 2004).

Our patient had absolute azoospermia. We decided to perform a testicular biopsy to increase the chances for retrieving viable sperm. Several authors have studied the success rate of testicular sperm retrieval in men with Y deletions (Brandell *et al.*, 1998; Silber *et al.*, 1998; Krausz *et al.*, 2000; Hopps *et al.*, 2003; Simoni *et al.*, 2004; Vogt, 2004). It has been shown in these studies that a proportion of patients with AZFc deletion may have sperm retrievable within the testis. Deletions involving the AZFb regions are generally associated with a poor prognosis for sperm retrieval. The probability of finding sperm in a patient with a complete AZFb deletion is virtually nil. However, a diagnosis of partial AZFb deletion is compatible with sperm retrieval. No sperm have been found in the testes of azoospermic men with AZFb + c deletions. We present here a new case with an unbalanced translocation t(Y;22) which generated the loss of the long arm of the Y chromosome involving a large part

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Table I. Genotype–phenotype correlation in adult males with Y-autosome translocation involving Yq11 region

Karyotype	Origin	Molecular analysis	Phenotype		References
			Sperm count	Testicular histology	
45,X male with Y-autosome translocation					
t(Y;14)(q11;p?)	*de novo*	Intervals 6 and 7 deleted	Azoospermia	Complete germinal aplasia	Turleau *et al.*, 1980; Andersson *et al.*, 1988
t(Y;15)(q11;p?)	*de novo*	Intervals 1–6 intact	Normal fertility	NP	Subrt and Blehova, 1974; Andersson *et al.*, 1988
t(Y;15)(q11.2;p12)	*de novo*	Intervals 6 and 7 deleted	Azoospermia	NP	Schempp *et al.*, 1985; Gal *et al.*, 1987
t(Y;22)(q11.1;p?)	*de novo*	Intervals 5, 6 and 7 deleted	Azoospermia	Only Sertoli cells	Arnemann *et al.*, 1991
t(Y;22)(q11.23;p11.2)	*de novo*	NP	Normal fertility	NP	Callen *et al.*, 1987
t(Y;22)(q11.2;q11.1)	*de novo*	AZFb and AZFc deleted	Azoospermia	Sperm seen on wet preparation Sertoli cell only on histology	Present case
Balanced reciprocal translocation					
46,X,t(Y;1)(q11.2;p34.3)	NP	No deletion of AZF regions	Cryptozoospermia	Degeneration of immature germ cells No mature spermatids	Pabst *et al.*, 2002
46,X,t(Y;1)(q11;q11)	Paternal	NP	Oligospermia	NP	Teyssier *et al.*, 1993
46,X,t(Y;3)(q11.2;q12)	*de novo*	NP	Azoospermia	Maturation arrest at the stage of primary spermatocyte	Gonzales *et al.*, 1981
46,X,t(Y;5)(q11.2;p15.3)	*de novo*	NP	Azoospermia	Maturation arrest at the stage of primary spermatocyte	Dutrillaux and Gueguen, 1975
46,X,t(Y;6)(q11.23 ~ q12;p15.3)	NP	Interval 6 intact	Oligozoospermia	Reduction of spermatogenesis	Delobel *et al.*, 1998
46,X,t(Y;7)(q11.2;p22)	NP	NP	Azoospermia	Spermatogenic arrest No sperm	Sasagawa *et al.*, 1993
46,X,t(Y;14)(q11;p11)	*de novo*	NP	Azoospermia	Maturation arrest at the stage of secundary spermatocyte	Laurent and Dutrillaux, 1976
46,X,t(Y;16)(q11;q13)	*de novo*	NP	Oligozoospermia	Partial block at spermatid formation Scanty sperm	Faed *et al.*, 1982
46,X,t(Y;16)(q11;p13)	*de novo*	NP	Azoospermia	Maturation arrest of spermatogenesis	Abeliovich *et al.*, 1986
46,X,t(Y;16)(q11.21;q24)	*de novo*	No deletion of AZF regions	Oligozoospermia	NP	Giltay *et al.*, 1998, 1999
46,X,t(Y;17)(q11.21;q12)	NP	NP	Azoospermia	Maturation arrest at the stage of secundary spermatocyte No sperm	Laurent *et al.*, 1982
46,X,t(Y;19)(q11;p or q13)	*de novo*	NP	Azoospermia	NP	Smith *et al.*, 1979
Unbalanced translocation					
46,XY,-14, + der(14)t(Y;14)(q11;p11)	NP	NP	Azoospermia	Complete arrest at the sperm head stage No sperm	Ratomponirina *et al.*, 1985
46,X,der(Y)t(Y;15)(q11.23;p11)	*de novo*	NP	Perspective sperm donor	NP	Bardoni *et al.*, 1991
46,X,der(Y)t(Y;15)(q11.23;p11)	Paternal	Interval 6 intact	Oligozoospermia	NP	Bardoni *et al.*, 1991

NP = not performed.

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of AZFb, AZFc and Yq heterochromatin regions and with sperm seen in the testis biopsy. However, our STS analysis is not sufficient to precisely characterize the extent of the deletion. The only way to demonstrate that a complete AZFb deletion has occurred in this specific case would be a sequencing approach since rearrangements interfering with AZFb region sequences cannot be excluded in a translocated Y chromosome. Unfortunately this is not available in our case.

The sperm could thereafter be used for ICSI in combination with preimplantation genetic diagnosis. The chromosomal risks of this translocation in offspring are aneuploidy of chromosome 22 and sex numerical anomalies. Nevertheless, the most important genetic risk for the couple concerns sex chromosome anomalies because monosomy or trisomy 22 are not viable.

In conclusion, our case highlights the importance of documenting (Y;autosome) translocations with molecular and testicular tissue analyses in order to establish a genotype–phenotype correlation.

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Submitted on September 16, 2004; resubmitted on March 22, 2005; accepted on March 31, 2005

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