

9 X

(2)

(1)

(1)

(1)

(2)

2009/02/18

2009/08/04

GTG-banding
(p22.2) X

(q31) 9

. 46, Y, t(X;9)(p22.2;q31)

(FISH) Fluorescence In Situ Hybridization

Microdeletion

Y

(AZF) Azoospermia factor

X

:

A Case Report of a Reciprocal Translocation Between Chromosome X and Chromosome 9 Found in Two Infertile Brothers Inherited from Their Mother

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ABSTRACT

During our genetic research to find association between genetic defects and idiopathic male infertility in Syria, we found amongst 160 patients studied, a rare reciprocal translocation between the short arm of chromosome X (p22.2) and the long arm of chromosome 9 (q31) in two infertile brothers. Our finding was confirmed by Fluorescence In Situ Hybridization (FISH), also a conventional cytogenetic study was done to their mother which revealed that this translocation is inherited from her. Microdeletion of the long arm of the Y chromosome was studied for both brothers too, but no microdeletion was found. These findings show clearly the important role of X-autosome translocation in causing Azoospermia and male infertility In male carriers.

Key words: Idiopathic male infertility, Azoospermia, Reciprocal translocation, Karyotype, Chromosome pairing.

2002) male infertility
 spermatogenic failure

infertility
 %30 .
 (WHO
 .Azoospermia

chromosome pairing

spermatogenic arrest

.(Mau-Holzmann 2005)

()

reciprocal translocation

Karyotype

27

()

FSH= 5.7, Testosterone=29.8,)

(LH=1.6, Prolactine= 8.2

primary insufficient

.spermatogenesis with early maturation arrest

25

()

()

:

Cyto) 37 72
 (Gen, Sinn, Germany
 Human 1995) GTG-banding
 20 .(chromosomes
 X 9 reciprocal translocation
 p22.2 X
 9
 : ISCN(2005) q31
 .(2) 46,Y, t (X;9) (p22.2; q31)

:(FISH) Fluorescence In Situ Hybridization

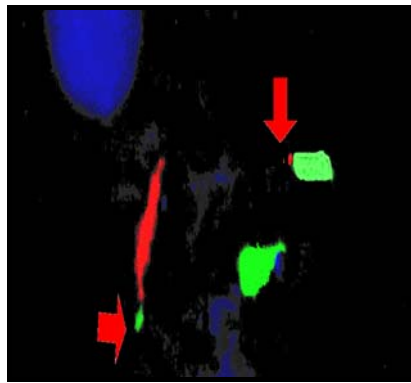
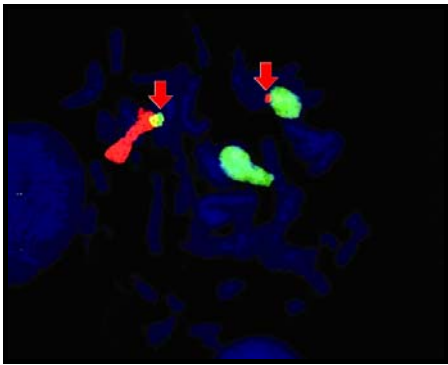
(FISH)

Fluorescein isothiocyanate 9
 (Chromosome paint XCP9 Meta System GMBH) .(FITC)
 Chromosome) (Texas Red) X
 : (paint XCPX Meta System GMBH
 (PH7) 2XSSC
 (5+ 5) 10 % 100 85 70
 C 3 75°C
 (PH7) 0.1 SSC 37°
 72° C (PH7) 0.4 SSC
 20 x SSC T
 DAPI 15

FISH

X
9

: ISCN (2005)
 .(1) 46, Y, t (X;9) (p22.2;q31) ish der(9) (wcpX⁺); der (X) (wcp9⁺)



(1)

X () ()
 9 (Texas Red) ()
 Fluorescein isothiocyanate (FITC)
 DAPI .X 9

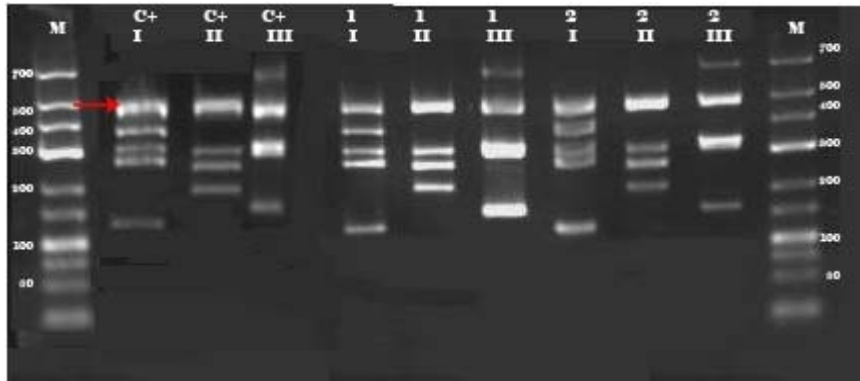
:
X

()
52

(2) 46, X, t (X;9) (p22.2;q31)

DNA (STS) sequence-tagged sites
 SRY internal control (ZFX/ZFY)
 Positive control (C⁺) (microdeletion AZF DNA)
 .% 2.5

Y AZF Y Y
 (3)



AZF I, II, III (3)
 internal control .%2.5
 .(472bp) SRY (495bp) ZFX/ZFY
 M C+
 .bp 25 700
 .C⁺ 2 1 AZF

reciprocal translocations
 (Hook and Hamerton, 1977)

.nonhomologous chromosomes

Kalz-Füller *et al.*, 1999.)

(Waters *et al.*, 2001

()
X
AZF
X
Sai Ma) X
(*et al.*, 2003
Jamieson *et*)
X (*al.*, 1996
spermatocytes
(Buggea , 2000)
chromosome pairing
spermatogenic arrest
()
(primary insufficient spermatogenesis with
.early maturation arrest)

9 X X
X
9

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