

Y

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A Case of Azoospermia Associated with Yq Deletion

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= Abstract =

Different Y mutation in Yq11 occurring de novo in sterile males were first described 19 years ago. Since the phenotype of the patients was always associated with azoospermia or severe oligospermia, it was postulated that these mutations interrupt a Y spermatogenesis locus in the euchromatic Y region(Yq11) called azoospermia factor(AZF). Recently, it became possible to map AZF mutations to different subregions in Yq11 by molecular deletion mapping. This indicated that azoospermia is possibly caused by more than one Y gene in Yq11 and the Yq11 chromatin structure. The frequency of AZF mutations in idiopathic sterile males(5 – 20%) may indicate a need for a general screening programme for its analysis in infertility clinic. We have experienced a case of deletion distal to Yq11 region in azoospermic patient. So we report this case with a brief review of literatures.

Key Words : Azoospermia, Yq11 deletion

Y (euchromatin)
(interstitial deletion)
, Yq11
Yq11
(microdeletion) 가
700 3 가
Yq11 Yq11 3
가
(azoospermia factor : AZF) a,b,c
Y
가 AZF 가
Y 11
31 1
가
175cm, 90kg, 120 / 80 mmHg, 36.5°C, 80 /
0 12cc
FSH : 21.0 mIU/mL, LH : 5.0 mIU/mL, prolactin :
12.0 ng/mL, testosterone : 4.13 ng/mL 3.0
cc, pH 8.0
가 가
, Leydig Y
11 가 (Fig. 1).
Clomiphene 가
(intracytoplasmic sperm injection : ICSI)

가 (testicular sperm extraction : TESE)

가

40 - 60%

(De Kretser, 1997).

Klinefelter , 46,XX , Kallmann , Kartagener

(congenital absence of the vas deferens) (cystic fibrosis) 가 Y

azoospermia : DAZ) RNA (deleted in 1(RNA binding motif 1 : RBM1) (Ma et al., 1993 ; Reyo et al., 1995).

가

가 가

3.65% . 가

가
(Chandley et al., 1972 ; Bourrouillou et al., 1985).

가
(Cummins and Jequier, 1994).
Y (deletion)
(Chandley and Cooke, 1994 ;
Reijo et al., 1995 ; Reijo et al., 1996). 가
50% 가
(Anguiano, 1992).

가 가 .
(Baschat, 1996).

가
(Vogt, 1995).

(chromatin)
가
(Evans et al., 1978).
Y (Tiepolo and
Zuffardi, 1976). Y
(azoospermia factor : AZF)가

Y
(microdeletion) (Camarri et al., 1993 ; Ma et al.,
1993 ; Reyo et al., 1995). YRRM1(Y chromosome RNA
recognition motif 1) RBM1 가
RNA (RBM) Y

RBM1 . DAZ 가

RBM1 DAZ RNA
 (transcription) . 가
 DAZ 가 가
 가
 가 370
 (Vogt et al., 1996). Yq11 3 가 AZF (locus)가
 AZFa AZFb
 RBM1 AZFc
 DAZ .
 가 Y
 (Vogt et al., 1996 ; Pryor et al., 1997).
 .
 Y
 (polymorphism) 가
 AZFa, AZFb, AZFc 가
 (de novo) .
 가 .
 가 .
 가 .
 . AZFa AZFb
 AZFc
 (Vogt et al., 1996). AZFc 가
 .
 (Kremer et al., 1997).

가 Y 11
 1 .

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