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**A Case of Successful Pregnancy in Patient with
Recurrent Spontaneous Abortion by Preimplantation
Genetic Diagnosis following IVF-ET**

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=Abstract=

It was reported that the etiologies of recurrent spontaneous abortion are immunologic factors, endocrinologic problems, anatomical abnormalities, genetic abnormalities, infection, and unexplained factors. Among those etiologic factors, genetic abnormalities occur in about 5% of the couples who experience recurrent spontaneous abortions, and most common parental chromosomal abnormality contributing to recurrent abortion is balanced translocation.

The advent of in vitro fertilization (IVF), the development of skills associated with the handling of human embryo, and an explosion of knowledge in molecular biology have opened the possibility of early diagnosis of genetic disease in preimplantation embryos. Therefore preimplantation genetic diagnosis (PGD) is indicated for couples, infertile or not, at risk of transmitting a genetic disease.

A case of successful pregnancy and term delivery by PGD using fluorescence in situ hybridization (FISH) technique in patient with recurrent spontaneous abortion due to balanced translocation is presented with brief review of literatures.

Key word : Recurrent spontaneous abortion, Balanced translocation, Preimplantation genetic diagnosis (PGD), Fluorescence in situ hybridization (FISH)

I.

가 3 2 , 0.4-1%
(Wilcox et al., 1988).
, (immunologic) 50%, (endocrinologic) 17%, (anatomical)
12%, (genetic) 5%, (infection) 5%, (unexplained) 10%
(Hill, 1996).
가 , ,
(balanced translocation)가 가 (Plouffe et al., 1992;
Tulppala et al., 1993).
(assisted reproductive technology, ART) (micromanipulation)
가
(preimplantation genetic diagnosis, PGD) 가 .
(polymerase chain reaction, PCR) fluorescence in situ hybridization(FISH)
, (sex) PCR FISH
(single gene defect) PCR . FISH
(Kearns and Pearson, 1994).
FISH .
(in vitro fertilization and embryo
transfer, IVF-ET) FISH PGD 1

II.

: O O, 28
:
: 0-0-3-0 1995 , 1996 7 , 8 , 6 .
: 15 30 5 ,
.
:
가 :
: 1994 1996 12
.
(hysterosalpingography)
(intrauterine adhesion) (submucosal myoma)
(prolactin) 15.1 ng/ml,
(thyroid stimulating hormone, TSH) 1.0 μ U/ml, (luteinizing hormone, LH) 14.2
mIU/ml, (follicle stimulating hormone, FSH) 5.7 mIU/ml, testosterone 0.16
ng/ml . antinuclear antibody(ANA) , lupus
anticoagulant(LAC) , anticardiolipin antibody(ACA) IgG/IgM , rheumatoid factor(RA)
<20 U/ml, antithyroglobulin antibody <20 μ /ml, antiTSH-receptor antibody 8.4%,
antimicrosome antibody <10 U/ml . 1996 12 16
 , 12 30
, (luteal phase defect)
 . 1997 1 3 . 46XY
46XX, t(11:12)(q22.3;q11.21) 가
 .
: 1997 2 17 gonadotropin-releasing hormone(GnRH)
agonist(Decapeptyl, D-Trp-6-LH, Ferring, Malmo, Sweden) (luteal
phase long protocol) (Neveu et al., 1987; Tan et al., 1992 & 1994).
human menopausal gonadotropin(hMG; Humegon, Organon, Holland) human follicle
stimulating hormone(hFSH; Metrodin, Serono, Switzerland)

(step-down fashion) . 3 15

10 (preovulatory phase oocyte)

FISH (polar body) (micromanipulator)

, 1 x 10⁵/ml 18

(pronucleus)

72-74 (denaturation) 70%, 85%,

100% , FISH probe WCP[®] Chromosome Patient

DNA FISH Probe(Vysis, Framingham, MA) 11 12 probe 1μl 7μl

hybridization buffer 1μl H₂O 74 5

45 slide warmer 10μl probe mix loading cover

slip 42 (humidity box) 12

. 50% formamide, 2X SCC(pH 7.0-8.0) 3 , 2X SCC(pH 7.0) 10 , 2X

SCC, 0.1% nondiet-P40 1 ,

4,6-diamino-2-phenylindole(DAPI) counter staining coverslip (Olympus,

Japan)

10 6 가 4 가

(Fig. 1) 3 3 18 , 4

4 1 2 , 8 1 2 .

progesterone in oil(Progest, Samil pharm., Korea) 50 mg

. 3 31 , 13 -hCG가 407 mIU/ml 4 15 6⁺⁴

(gestational sac)

(crown-rump length, CRL) 3.1 mm 6

11 11 40⁺⁶

3525 gm

a. normal

b. balanced translocation

Fig. 1. Fluorochrome-labeled chromosome 11 (red) and 12 (green)- specific DNA
by in situ hybridization

.

가 가 ,
가 3-8% 가
(Plouffe et al., 1992; Tulppala et al., 1993).
(mental retardation)

가 , (chorionic villi
sampling, CVS) (amniocentesis)
가

intracytoplasmic sperm injection (ICSI) 가
가 ,
가

가 , (cleavage
stage) (blastomere biopsy), trophectoderm (Dokras et al.,
1990; Verlinsky et al., 1990; Delhanty, 1994). 가
1 (Verlinsky et al., 1990), 1
1 가 1 (first meiotic division)

(zona pellucida) (assisted hatching) 가
(Cohen et al., 1990)

(Krzyminska et al., 1990; Hardy et al., 1990).

8- 가 가
 , 가 (high mitotic index)
 (compensation) 가 (Krzyminska et al., 1990), , 가 (survival rate)
 (Hardy et al., 1990), , structural junction 가
 (Dale et al., 1991), 1-3
 , mosaicism .
 PCR FISH가
 . PCR (mutation)
 (Grifo et al., 1992; Muggleton-Harris et al., 1993),
 가 가 . PCR
 (DNA sequence)
 가
 가 FISH
 , FISH (DNA
 probe) (aneuploidy)
 (polyploidy) 가 (Griffin et al., 1992; Munne et al., 1993; Verlinsky et al., 1996).
 (metaphase) (interphase) 가
 (Hopman and Ramaekers, 1988).
 (deletion), , (insertion) 가 .
 가 ,
 (X-linked disease)
 FISH

.

FISH

1

.

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