하나 SPA가 진행되는 동안에 HP에 의한 막지질 과산화 등의 부정적 영향이 크지 않아 SPA 결과에 크게 저하시키지 않는 것으로 사료된다. 또한 SA와 HP는 SPA 결과를 저하시키는데 상승적효과를 보이는 것으로 사료된다.

P-4

Association of Reproductive Abnormalities with Pericentric Inversion of Chromosome 9

Department of Obstetrics and Gynecology, Genetic Research Laboratory*, Samsung Cheil Hospital & Women's Healthcare Center

Hyun Mee Ryu, Jin Mi Kim*, Young Mi Kim*, So Yeon Park*, Soo Kyung Choi*, Inn Soo Kang

Pericentreic inversion of the chromosome 9, inv(9)(p11;q13), is occurs commonly with an incidence of 1% to 1.65% and some cytogeneticists would consider it as a normal variant. This entity is categorised as a minor chromosomal rearrangement with normal phenotypes. However, many reports in the literature suggested that it may be associated with subfertility, recurrent abortions or other chromosomal abnormalities arising as a result of having inv(9). Herein, we analyzed the incidence and clinical significance of inv(9) among those who had reproductive abnormalities. Peripheral blood karyotypes(2070 cases) performed due to recurrent spontaneous abortions or infertility in Samsung Cheil Hospital over the last six-years were selected. Cases for prenatal genetic diagnosis were excluded from this study. Thirty one cases from 30 couples had inv(9)(p11;q13) which gave an incidence of 1.5%. One case of inv(9)(p12;q12) was found and excuded from the statistics. There were two cases with 47, XXY, inv(9). Among 30 cases with inv(9), 22 couples(73.3%) had history of more than two spontaneous abortions. Five families(16.7%) had the problem of infertility. Three couples(10%) previously had babies with major congenital anomaly. Interestingly, one case had homologous inversion 9 but she was phenotypically normal. Although there may be a selective bias because relatively older subfertile couples were included retrospectively, these data suggest that inv(9) may often cause clinical problems such as recurrent spontaneous abortions, subfertility or chromosomal abnormality in the offspring of the carriers.

P-5

Applications of PCR and Prins for the Sexing in Bovine Preimplantation Embryos

Departments of Animal Science and Genetic Engineering, Korea Univ., Seoul 136-701*

Seung Woon Seo, Hong Jun Lee, Ki Dong Kim, Sung Soo Park* and Sang Ho Lee

Predetermination of sex in preimplantation embryos is of great value in the domestic animalproduction and clinical medicine since it provides a means to select animal sex of interest and diagnosis the human genetic diseases prior to implantation. Polymerase chain reaction (PCR) and primed *in situ* DNA synthesis (PRINS) were used to determine the sex of *in vitro* fertilized bovine embryos. Eight-cell to morulae stage embryos were obtained from *in vitro* fertilization (IVF) and subsequent coculture with oviductal epithelial

cells. DNA was prepared by suspending single embryos in PCR lysis buffer containing 200μg/㎖ proteinase K and incubated at 50℃ for 1hr. Metaphase chromosome spreads were prepared from nocodazole-treated embryos by air-drying method. To eliminate possible false positive signals, two sets of bovine-and Y chromosome-specific primers were used in the PCR. Two amplified products (bovine- and Y-specific) were obtained in male samples whereas only one product (bovine-specific) in female. FISH and PRINS were used to identify the Y chromosome on metaphase spreads. The fluorescent Y-specific signal was stronger in PRINS than in FISH.

The results suggest that a rapid, accurate and efficient sexing is now possible in bovine preimplantation embryos produced *in vitro* using PCR. This was evidenced by PRINS.

P-6

Single Cell Analyses of Dystrophin Gene and Sexing Using Whole Genome Amplification

Genetic Research Lab., Dep. of Obstetrics and Gynecology[1],
Sumsung Cheil Hospital & Women's Healthcare Center,

Soo Kyung Choi, Jin woo Kim, Eun Hee Cho, So Yeon Park, Hyun Mee Ryu[1], Inn Soo Kang[1]

Recently, developed primer extention preamplification(PEP) method amplifies the whole genome and simultaneous multiple DNA analyses became possible. It leads to the development of preimplantation genetic disease diagnosis using single cells from early embryo, sperm, polar body and oocyte. The most important advantage is the ability which can investigate several loci simultaneously and confirm results by analysing multiple aliquots for each locus. Whole genome from each single cells was amplified using 15-base oligonucleotide random primers. In this study, we performed PEP-PCR for applicate on prenatal and preimplantational genetic diagnosis in 20 cases of single amniocytes and 20 cases of single chorionic villi cells. We stuied 7 gene loci simutaneous analysis of single cells at two locus of exon 46, 47 and two VNTR(variable number tandem repeat) markers using 5'dysIII, 3'CA related to dystrophin gene and ZFY, alphoid repeat Y, DYS14 regions on chromosome Y. In all these cases, ninety seven percent of PEP reactions with single amniocytes and chorionic villus cells were successful. We obtained 38/40(95%) of accurate gender determination by comparing chromosome analysis and general PCR from gDNA. Therefore, these results have significant implications for a sperm or oocyte typing, prenatal and a preimplantational genetic diagnosis.

Group 2, discussion : 14:00~14:30

P-7

실패하였던 정관부고환문합술과 정관정관문합술에서 MESA-ICSI와 TESE-ICSI의 효용성

삼성제일병원 비뇨기과, 불임연구실[1]

김종현, 서주태, 박용석[1], 이유식

연구목적 : 폐쇄성 무정자증의 치료방법인 정관부고환문합술과 정관수술후의 정관복원을 위한 정관정관문합술을 시행후 실패하였던 경우, 재수술을 시행할 때 수술의 성공에 대하여 절대적인 확신을 가질 수 없다. 그러므로 재수술을 할 때 부고환이나 고환에서 정자를 채취하여 난자의 세포질내로