

Recurrent spontaneous abortion in IVF treatment

---A case report of mosaic turner's syndrome

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INTRODUCTION: Chromosome analysis is one of the basic examinations for evaluating the habitual abortion. Current chromosome analysis was performed by reading the karyotype of 20 cells. However, some mosaic pattern of chromosome abnormally might be misdiagnosed by reading this limited amount of cells. We present a case who suffered from consecutive spontaneous abortion for 5 times. Traditional chromosome study by reading 20 cells revealed a normal female karyotype 46 XX. While extending the cell number for analysis to 115, we found 5% of these cells were 45 X0. FISH (fluorescence in situ hybridization) examination was arranged and five cells with single-X gene were found in total five hundred cells. Accordingly, we suggest more detailed and advanced chromosome analysis when dealing with habitual abortion or unexplained infertility.

CASE REPORT: This 38 y/o, G3P0SA3 female visited our OPD due to recurrent abortion 1995. Her previous survey for habitual abortion included a normal karyotype of both her and her husband. Normal hysteroscopy finding, negative antiphospholipid antibody and negative anticardiolipin antibody. She was neither a case of SLE. There was neither no matching of HLA typing between her and her husband. She visited our center due to the long lasting secondary infertility and asked for ART (assistant reproductive technique) treatment. Three cycles AIH (Assistant insemination husband) were performed first without good result. She then received IVF-ET (in-vitro fertilization with embryo transfer) and one cycle FET (Frozen embryo transfer).

During her treatment, spontaneous abortion happened again twice even under the sufficient luteal phase progesterone support. Therefore, another chromosome analysis was suggested and the cell number for study was extended to 115 in this examination. Surprisingly, we found 5% of these cells were 45 X0. FISH examination was arranged and 500 cells were further studied. Among which, we found five cells with single-X gene. The final report of chromosome study was changed from normal 46 XX to mosaic turner syndrome.

She finally got pregnant through the oocyte donation program. One fetus with acrania was found by ultrasound at the 15 weeks of gestational age. The patient received cesarean section at the 34th weeks of pregnancy due to PIH (pregnancy induced hypertension). One fetus with multiple anomalies including acrania, brain adhesion to placenta, cleft lip and omphalocele was delivered. The other two fetuses were in normal range.

DISCUSSION: Chromosome anomaly is one of the leading causes of recurrent abortion. Our experience from this case revealed that traditional 20 cells chromosome analysis might miss diagnosis the mosaic form of chromosome anomaly. Accordingly, we suggested more advanced and detailed chromosome analysis in all cases with habitual abortion or unexplained infertility.