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Case Report

Successful pregnancy outcome in a patient with Robertsonian translocation (13; 14) (Q10:Q10) with recurrent pregnancy loss

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ABSTRACT

Robertsonian translocations are unique type of whole-arm translocation that result from 'centric fusion' of the long arms of two acrocentric chromosomes with loss of the short arms, thus reducing the number of chromosomes by one. RT's are present in 0.1% of the general population and 1% of the infertile population. Most frequent type of RT includes translocation rob (13; 14), whereas translocation rob (13; 15) and rob (14; 15) are rare. In the present report, RT in a female with spontaneous repeated abortions and infertility is reported. Cytogenetic analysis of a couple with repeated abortions revealed the presence of 45, XX, rob (13; 14) (q10; q10) chromosomal constitution in the female partner. The patient conceived after diagnostic laparoscopy followed by ovulation induction and intra uterine insemination and with proper antenatal care and support she delivered a healthy male baby with normal karyotype. The history of repeated abortions and infertility could be the outcome of unbalanced gametes (either monosomy or trisomy) resulting during the meiotic segregation of the balanced heterozygote female carrier. Cytogenetic analysis should be offered to all couples with unexplained recurrent abortions to evaluate the probable presence of any chromosomal aberrations.

Keywords: Recurrent pregnancy loss, Genetic cause, Robertsonian translocation

INTRODUCTION

Robertsonian translocations are unique types of whole-arm translocations that result from 'centric fusion' of the long arms of two acrocentric chromosomes with loss of the short arms, thus reducing the number of chromosomes by one.[1] It is mainly observed in 13, 14, 15, 21 and 22 chromosomes. Most frequent type of RT includes translocation rob (13;14), whereas translocation rob (13;15) and rob (14;15) are rare structural rearrangements.[2]

Robertsonian translocation is associated with various risks such as infertility due to unbalanced gametes, repeated pregnancy loss and cancers (acute and chronic myelogenous leukemia). It is the most common structural chromosomal abnormalities with an incidence of 0.1% of general population, 1.1% in couples with recurrent pregnancy loss and 1% of the infertile couples.[3]

CASE REPORT

A 26 year old lady married for the past 5 years, with history of repeated miscarriages and inability to conceive for 2 years reported to the department of reproductive biology of the Indira Gandhi Institute of Medical Sciences, Patna, India in March 2013.prior to this she had three miscarriages at 11 weeks, 8 weeks and 6 weeks.

On physical examination no abnormality was detected in both partners. All routine investigations were in normal limits. Lupus anticoagulant and anticardiolipin was

negative. Husband semen analysis was also in normal limits.

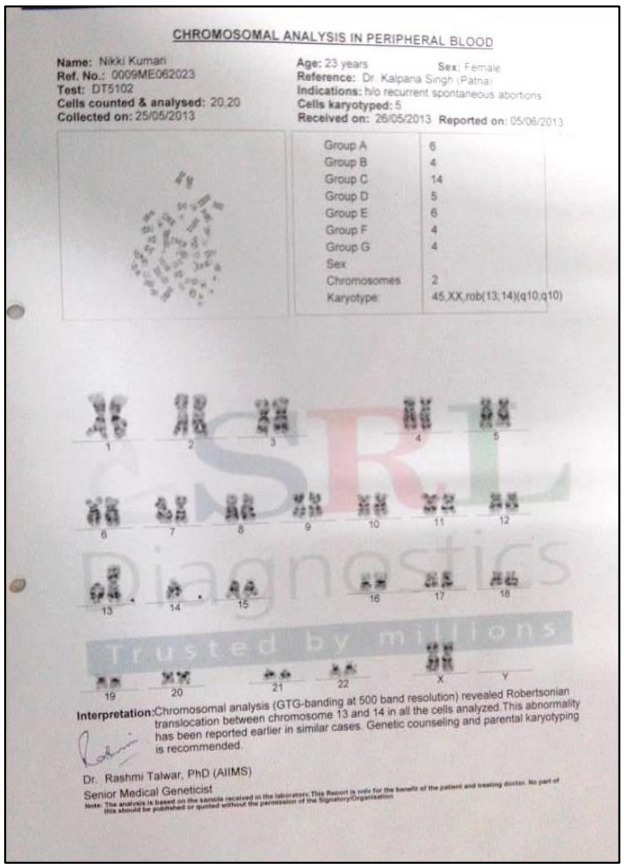

Figure 1: Chromosomal analysis in peripheral blood.

Cytogenetic analysis revealed an abnormality in the female partner with 45, XX, rob (13;14) (q10;q10) chromosomal constitution while the male partner showed normal 46, XY karyotype.

Diagnostic laproscopy with chromo-pertubation was done. Uterus, both ovaries and tubes were healthy. Both the tubes were found patent. Ovulation induction followed by intrauterine insemination was done. She then conceived in third cycle. Routine antenatal care with tender love and support was given.

She delivered a live male child by LSCS in March 2014 at 37 week gestation. Cytogenetic analysis of baby showed normal male karyotype;46XY.

DISCUSSION

Around 15 to 20% of all pregnancies in humans end in spontaneous abortions and 60 % of all spontaneous abortions in early pregnancies are a result of chromosomal aberrations during embryogenesis.[4] Although the cause is unknown in many instances, but parental chromosomal abnormality is one of the possible causes for recurrent first trimester miscarriages.[5] Evaluation of patients with a history of repeated spontaneous abortions requires careful consideration of genetic, anatomic, endocrine, infectious, and immunological factors. Assigning proper etiological role to each of these contributing factors is often unclear, however the specific information about the cytogenetic makeup of the couples and if possible of the abortus, still remains a primary focus during evaluation of such cases. The majority of pregnancy losses or neonatal deaths are reported to result by numerical chromosomal abnormalities especially trisomies of chromosome 13, 18, 21 etc. The structural aberrations can also be the cause of pregnancy loss and infertility. Robertsonian translocations are the commonest structural aberrations of the acrocentric chromosomes, the most common are the non-homologous forms i.e., those involving two different acrocentric chromosomes - either two different D group chromosomes (13, 14 and 15) or G group chromosomes (21 and 22) or a combination of D group and a G group chromosomes.[6] Studies indicate that when the Robertsonian translocation is maternal, there is greater risk that the fetus will exhibit an unbalanced phenotype.[7]

In this case report a non-homologous Robertsonian translocation between two D group chromosomes (13 and 14) in a female with repeated abortions is presented. The cytogenetic analysis revealed 45, XX, rob (13;14) (q10;q10) chromosome constitution in the female indicating the possible association of such anomaly with repeated abortions.

Balanced carriers of Robertsonian translocations are phenotypically normal. Almost all balanced carriers are heterozygous for the translocation and usually experience poor outcomes from pregnancy; they are at increased risk for spontaneous abortions and chromosomally unbalanced offspring. The main concern for people with a Robertsonian translocation is that they may have a child with extra genetic material, which can cause medical problems. For each pregnancy, the outcome depends on whether the sperm or the egg from the parent who has the Robertsonian translocation contains the Robertsonian translocation and/or normal chromosomes.

During pachytene stage in meiosis, homologous pairing of Robertsonian translocation is achieved by the formation of a trivalent. If alternate segregation occurs, then all gametes are potentially viable with balanced chromosomes. But, the adjacent segregation results in gametes nullisomic or disomic for one of the chromosomes involved in the rearrangement and consequently a zygote with trisomy or monosomy for one of the chromosomes involved.[8]

If the mother has a Robertsonian translocation involving chromosomes 13 and 14 and the father has normal chromosomes, there are different possibilities of segregation of gametes and outcomes of pregnancy. However, it is impossible to predict how often a certain pattern will be passed on to the offspring but it is quite

possible for a person who carries a balanced RT to have healthy children and many do. The offspring can inherit

- Neither of the chromosomes from the mother that were involved in the Robertsonian translocation. This will result in a normal healthy baby as they have inherited a normal set of chromosomes as in this case report.
- Both chromosomes from the mother that were involved in the Robertsonian translocation and normal copies of the chromosomes from the mother. This means that the baby has extra genetic material (i.e. the translocation is now unbalanced). This is likely to result in physical or mental disability. The type and severity of the disability depends on the extra chromosome. If the baby has an extra copy of chromosome 14, making 3 copies instead of 2 (Trisomy 14), the pregnancy is likely to end in an early miscarriage. If the baby has 3 copies of chromosome 13 (Trisomy 13, Patau's Syndrome), may born alive but they usually die in the first few weeks or months of life) whereas, zygotes with monosomy are not compatible with life.
- Both chromosomes from the mother that were involved in the Robertsonian translocation. This does not usually cause any medical problems, but the baby will be a carrier of the Robertsonian translocation just like their mother.

Prenatal diagnosis has been available to carriers of Robertsonian translocations for many years. Such couples must be counseled that they could opt for chorionic villi sampling or consider PGD in conjunction with assisted conception using IVF or intra cytoplasm sperm injection (ICSI). Thus cytogenetic analysis can help to solve the mystery of a recurrent miscarriage and also suggest remedial measures.

CONCLUSION

Balanced carriers of Robertsonian translocation have an increased risk for infertility, miscarriages and chromosomally unbalanced offspring with multiple congenital abnormalities and intellectual impairment. Cytogenetic analysis is a valuable tool for the reproducing couples with more than two spontaneous abortions to delineate chromosomal aberrations, if any. The early detection of chromosomal aberration helps for appropriate genetic counseling and allows parents to make an informed reproductive decision on subsequent pregnancies. Prenatal diagnosis offered to these couples on future pregnancies enables one to prevent social stigma of repeated abortions and implications societal barriers.

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REFERENCES

1. Gilgenkrantz S. Robertsonian translocations and abnormal phenotypes. Groupe de Cytogénéticiens Français. Ann Genet. 1989;32(1):5-9.
2. Jacobs PA. Mutation rates of structural chromosome rearrangements in man. Am J Hum Genet. 1981;33(1):44-54.
3. Fryns JP, Van Buggenhout G. Structural chromosome rearrangements in couples with recurrent fetal wastage. Eur J ObstetGynecol. 1998;81:171-6.
4. Garcia -Enguidanrs A, Calle ME, Valero J, Luna S, Domínguez-Rojas V. Risk factors in miscarriage a review. Eur J Obstet Gynecol Reprod Biol. 2002;102(2):111-9.
5. Sullivan AE, Silver RM, LaCoursiere DY, Porter TF, Branch DW. Recurrent fetal aneuploidy and recurrent miscarriage. Obstet Gynecol. 2004;104(4):784-8.
6. Kim S, Shaffer CG. Robertsonian translocations: mechanisms of formation, aneuploidy and uniparentaldisomy and diagnostic considerations. Genet Test. 2002;6:163-8.
7. Boue A, Gallano P. A collaborative study of the segregation of inherited chromosome structural rearrangements in 1356 prenatal diagnoses. Prenat. Diagn. 1984;4:45-67.
8. Gardner RJM, Sutherland GR. Chromosome abnormalities and genetic counseling. 4th ed. Oxford: Oxford University Press; 2004:122-137.

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