

Successful Pregnancy in a Woman with Recurrent Pregnancy Loss and Balanced Robertsonian Translocation: A Case Report

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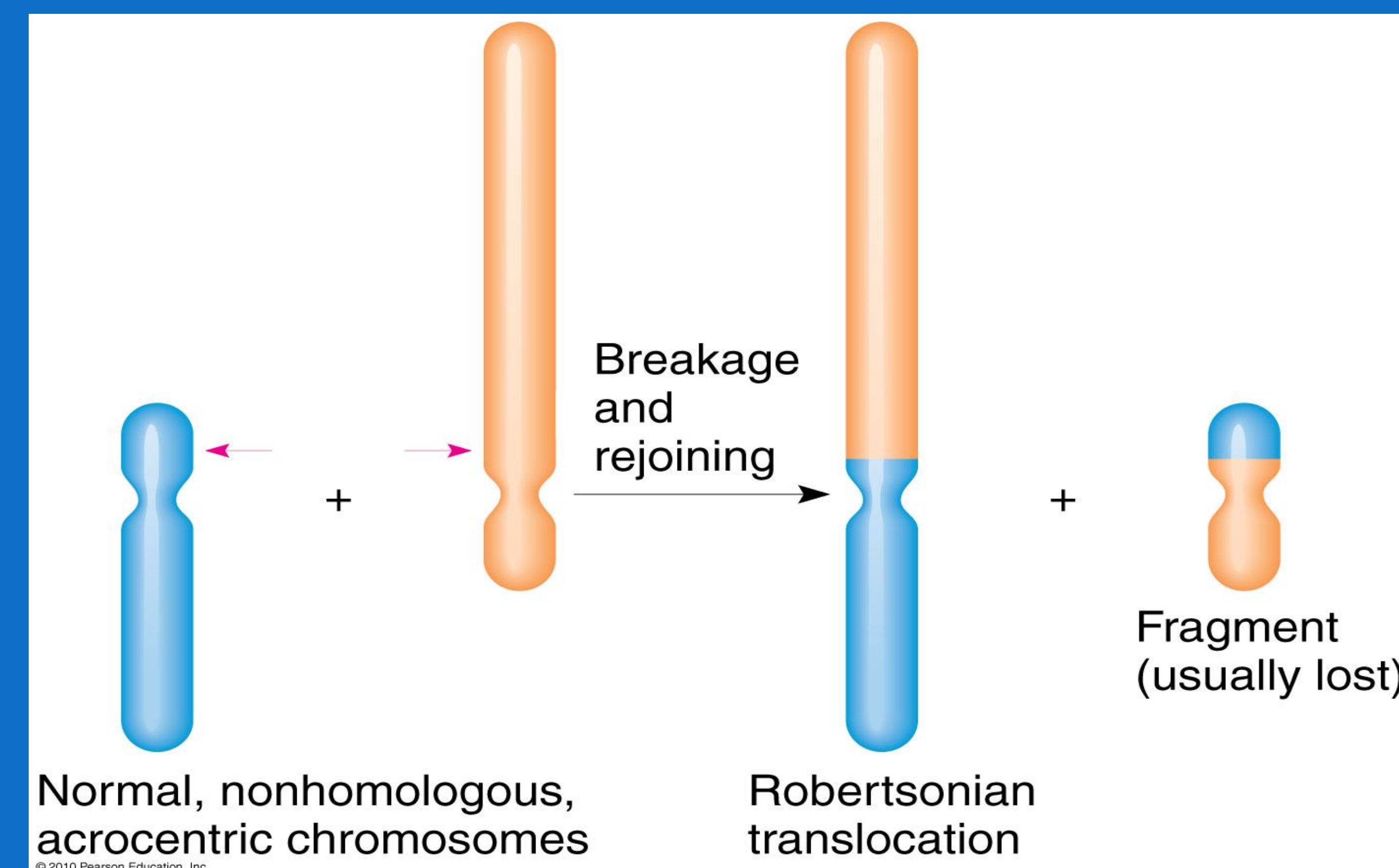
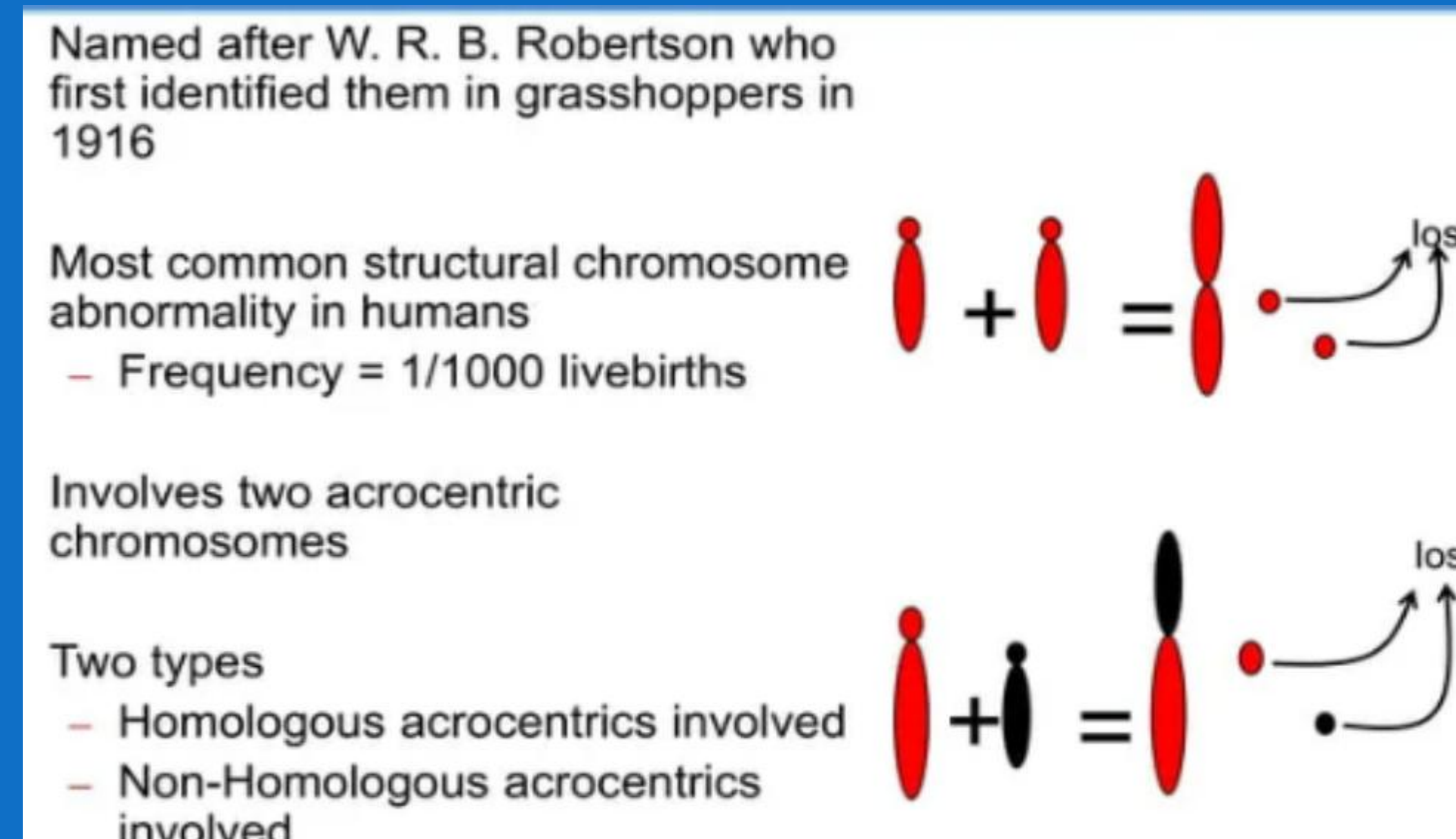
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Background: Balanced Robertsonian translocations (RT) are the most common chromosomal rearrangement responsible for subfertility and recurrent pregnancy loss, which occur in 3-5 % of couples. These balanced translocations result in infertility due to unbalanced gametes, and recurrent spontaneous abortions because of monosomies and trisomies or chromosomally abnormal embryos [4,5]. RT is mainly seen in chromosomes 13,14,15, and 21, but the most frequent variety involves translocations between chromosomes 13 and 14 (ROB13;14), while Rob13;15 and Rob,15;21 are rare types of chromosomal rearrangements. We present a case of a woman with repeated miscarriages who was found to be a carrier of balanced Robertsonian translocation on karyotyping, who had a successful pregnancy outcome after in vitro fertilization. Genetic testing of the conceptus and karyotyping of the parents are important steps in the management of recurrent pregnancy loss.

The case: Mrs A, a 26-year-old woman, initially presented with anovulatory infertility as a result of polycystic ovarian syndrome. After routine and special investigations and a normal semen analysis, she underwent several cycles of ovulation induction and conceived three times but had three consecutive miscarriages. All three times, intrauterine pregnancy was confirmed and cardiac activity was demonstrated at 6-7 weeks by USG, but she miscarried the pregnancy subsequently.

Patient was investigated for APLA and infections, and her laboratory reports were negative for both. She was offered genetic counselling and testing after the third pregnancy loss, but the patient deferred and wished to try one more time. After the fourth abortion, the abortus was sent for chromosomal analysis. The result of the karyotype was abnormal (trisomy), following which both husband and wife were subjected to karyotyping.

The woman had a balanced Robertsonian translocation. Cytogenetic analysis revealed an abnormality in the female partner with 45, XX, Robertsonian (13;14) (q10; q10) chromosomal constitution, while the male partner showed a normal 46, XY karyotype. After the karyotype, the woman was suggested for IVF. The patient underwent an IVF cycle with a donor egg and conceived in the first cycle. This resulted in a successful twin pregnancy. The course of pregnancy was uneventful, and she had routine antenatal care. The woman gave birth to twin boys by vaginal delivery after 10 years of subfertility. Both babies were healthy with 2410 grams and 2910 grams birthweight.



Robertsonian translocation at 13,14 chromosome

Discussion: First-trimester pregnancy losses occur most frequently due to chromosomal abnormalities and account for 50% of abortions [1]. Of these about 52 % are trisomies, 29% are monosomies (45X), 16 % are triploidites and 6 % tetraploids. Out of these, 4% are structural rearrangements.

Our patient had an abnormal karyotype of the conceptus, and the woman had a balanced Robertsonian translocation. Parental Chromosomal abnormalities are the most common cause of repeated first-trimester pregnancy losses and account for 3.5% of couples as opposed to .7% in the general population. Robertsonian balanced translocations are the commonest structural rearrangements found in couples with RPL. Mohammad Hasanazadeh-Nazar Abadi and colleagues in their study of RPL found six out of ten related members of a single family with RPL had balanced translocations; in these couples, there was a history of consanguineous marriages [2].

Common translocations occur between chromosomes 13, and 14, followed by 13, or 14, and 21. Our patient had a (13,14) (q10; q10) chromosomal constitution, while her husband had a normal karyotype. Filiz Ozen et al report a case of RT (14,15) in a male parent, and the female had a normal (46 XX) karyotype in a couple with habitual abortion [3].

Once diagnosed, managing these individuals is a challenge, although favourable pregnancy outcomes have been observed, it is a chance occurrence. Many carriers of 'Balanced Reciprocal translocations' may require ART with in vitro fertilization to conceive.

Conclusion: Genetic testing of the conceptus and karyotyping of the parents is an important step in the management of recurrent pregnancy loss. In Vitro Fertilization, with donor egg, may be a viable option for a successful pregnancy in women with Robertsonian translocation

References

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