A Letter
Open Access

Novel Balanced Translocation (4;20) with Recurrent Spontaneous Abortion--A Case Report
Salil Vaniawala, Pankaj Gadhia
Molecular Cytogenetic Unit, S. N. Gene Laboratory and Research Centre President Plaza – A, Near RTO Circle, Surat, India
Corresponding author email: pankajkgadhia@gmail.com
International Journal of Clinical Case Reports, 2016, Vol.6, No.9 doi: 10.5376/ijccr.2016.06.0009
Received: 02 Nov., 2015
Accepted: 13 Dec., 2015
Published: 19 Feb., 2016
Copyright © 2016 Salil Vaniawala, and Pankaj Gadhia, This is an open access article published under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.
Preferred citation for this article: Salil Vaniawala, and Pankaj Gadhia, 2016, Novel Balanced Translocation (4;20) with Recurrent Spontaneous Abortion--A Case Report, International Journal of Clinical Case Reports, 6(9) 1–2 (doi: 10.5376/ijccr.2016.06.0009)

Abstract
Chromosomal abnormalities are one of the major causes of infertility in man and woman. A couple with history of recurrent spontaneous abortion (RSA) was referred to our laboratory for chromosomal analysis. The analysis revealed normal 46,XY karyotype in male and a novel balanced translocation 46,XXt(4;20)(q11;p13) in female partner. This translocation with breakpoints has not been reported in the literature particularly with RSA.

Keywords RSA; Balanced translocation; Chromosomal anomalies; GTG banding

Introduction
The recurrent spontaneous abortion (RSA) is known complication of pregnancy. The cause is multifactorial and associated with parental age, infectious diseases, congenital anomalies and genetic abnormalities (Dudley and Brauch, 1989). RSA is historically defined as three or more consecutive pregnancy losses before 20 to 22 weeks of gestation (Stirrat, 1990).

Balanced translocations are frequent structural chromosomal rearrangements observed in humans. The usual translocations reported are between X chromosome and autosomes. The most frequently chromosomes involved are 4, 6, 9, 12, 15 and 18 (Tullu et al., 2001).

Around 15 to 20% of all pregnancies in human end up with spontaneous abortions (SABs).

The incidence of chromosomal abnormalities in those abortions is around 50%. Although the causes is unknown but parental chromosomal anomalies is one of the possible cause for RSA.

Case report
A 28-year old female patient reported to our cytogenetic laboratory with history of three times recurrent spontaneous abortions. There was no bad obstetrics history in family. Her height was normal 154 cm and weight was 58 kg having no consanguinity. She had well developed secondary sexual characters.

Chromosomal analysis was conducted on heparinized blood samples of both male and female by PHA-stimulated blood cultures. Chromosome preparation were subjected to GTG-banding (Seabright, 1971) and preparation of karyotyped was done according to ISCN (2009).

The results showed balanced translocation between long arm of chromosome 4 and short arm of chromosome 20. A karyotype of 46,XX,t(4;20)(q11;p13) was revealed (Figure 1) with clinically normal looking female phenotype, while male partner has a normal genotype with 46,XY.

Figure 1 G-banded karyotype of the female patient showing 46,XX t(4;20)(q11;p13)
Discussion

Cytogenetic anomalies have known to cause male and female infertility, but the exact mechanism by which how chromosomal anomalies induce infertility is yet to be proved. Several reports indicated that the chromosomal abnormalities which ranges from 2.2% to 15.7% for infertile men and an around 15 to 20% of all pregnancies in human end in spontaneous abortions (De et al., 2015).

In the current case report, we present a family of which a male apparently healthy with 46,XY chromosome complement while female has three recurrent spontaneous abortions (RSA). A karyotype of female revealed a balanced translocation t(4;20) with break points of 4q11 and 20p13. To best of our knowledge balanced translocation t(4;20) with break points (q11:p13) is a novel one and completely different from one reported by Nigam et al., (2014).

They have revealed balanced t(4;20) (q12;q13.1) with absence of endometrium and primary amenorrhea. In our study, we found presence of endometrium without primary amenorrhea instead she had three times RSA and her phenotype was completely normal. Therefore, we suggest that such novel cases with normal phenotype be reported for better understanding of involving autosomal balanced chromosomal abnormalities in infertility.

Conclusion

The present study reported a novel balanced translocation [t(4;20)] could be one of the reasons for a recurrent spontaneous abortion. Hence, cytogenetic analysis should be mandatory for all couples with reproductive failures along with adequate genetic counselling to make informed decision regarding subsequent pregnancies.

Ethical clearance

The ethical clearance was obtained from Institutional ethical committee.

Author’s contribution

The work was carried out in collaboration. Author PG designed study, wrote protocol and prepared first draft of MS. Author SV performed culture and microscopic analysis and made preparation of karyotypes. PG managed the literature searches and analysis of data. Both authors read and approved the final MS.

References


http://dx.doi.org/10.4103/0974-1208.158623


ISCN, 2009, An International system for human cytogenetic nomenclature


http://dx.doi.org/10.4103/0974-1208.130867

Sea bright M., 1971, A rapid banding technique for human chromosome, Lancet, 2: 971-972


http://dx.doi.org/10.1016/0140-6736(90)92159-F