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Chromosome (8) pericentric inversion and recurrent pregnancy loss: a case study

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Abstract

Recurrent pregnancy loss (RPL) or Recurrent miscarriage (RM) can be defined as three or more consecutive pregnancy losses before 24 weeks of gestation. Many studies have proven that parental chromosomal abnormalities represent an important etiology of RM. This case of study was run to identify the role of chromosome rearrangement in RPL (1). To achieve the aim, a case of a phenotypically and normal physical examinations healthy couple, were subjected to cytogenetic analysis apparently after experiencing three RPLs. In their past the investigations were only limited to physical, imaging and routine laboratory testing in which no abnormal values were ever reported. Herein, the cytogenetic tests using G-banding showed that the wife was a carrier of pericentric inversion on chromosome 8; precisely 46,XX inv (8) (p11;q23), estimating the possible origin of the multiple pregnancy loss in this case. This finding underlies the importance of cytogenetic investigations in the routine management of RPL.

I. Introduction

In general; recurrent pregnancy loss (RPL) is one of the most challenging reproductive issue that affects couples worldwide. Three or more consecutive early pregnancy losses is considered RPL. Up to 50% of the time, the cause of recurrent pregnancy loss is unknown (1). Recurrent pregnancy loss can be classified into two basic categories: primary RPL, which occurs when a woman has never given birth to a live child, and secondary RPL, which occurs when a woman has previously given birth to a live child (2). However, Chromosomal abnormality in either parent is considered to be the one of the leading cause of recurrent spontaneous miscarriages (3).

This article's major focus will be on a case of a phenotypically healthy couple who experienced RPLs. Their Physical Examination showed no physical abnormalities, meanwhile, the principal laboratory testing did not report any abnormal values. Cytogenetic tests were carried out using G- banding and showed that the wife is a carrier of pericentric inversion on chromosome 8.

Etiology: The following broad categories can be used to classify the reasons for recurrent pregnancy loss (RPL): Antiphospholipid antibody syndrome, genetic, anatomic, endocrine, immune, and environmental factors (1). As part of the genetic role, RPLs were connected to chromosome numerical and structural abnormalities. 2–8% of cases possess a connection to particular parental chromosomal rearrangements, including balanced translocation and inversion (2). Both pericentric and paracentric inversions, which are internal rearrangements in which the centromere is included in the former but not in the latter, can result in irregular distribution of chromosomal material in gametes during meiosis (4).

Case presentation: A 27-year-old Libyan female was referred to the reproductive specialists with concerns about recurrent pregnancy loss. The patient experienced 2 unexplained miscarriages in the 15 months of marriage, the first occurred at 12 weeks gestation and the other at 13 weeks. The third miscarriage was reported after two years of marriage at about 12 weeks gestation. In their late third year of marriage, an Ultrasound examination revealed 7 weeks gestation of a single pregnancy with cardiac activity; 6 weeks later the pregnancy was lost. Following the loss of the third pregnancy, the couple underwent several investigations, and their clinical and obstetric history was carefully recorded. The age of the spouse, the number of RPLs, and the potential for additional abortion causes, such as uterine anomalies, hormone deficiency, and history of any clotting disorder or autoimmune disease were also investigated. Part form irregular menstrual cycles which the wife experienced after her second pregnancy loss no complications were reported in the partners. Furthermore laparoscopic revealed no abnormalities and no evidence of endometriosis.

Cytogenetic analysis: Lymphocyte karyotype using the G-banding technique (figure. 1) revealed that the wife carries a pericentric inversion of chromosome 8, inv (8) (p11; q23) whereas the husband showed a normal genotype (46, XY). Unfortunately, the aborted fetus or

the pregnancy wastage was not available for karyotyping. However, in this particular inversion involving chromosome 8, both male and female carriers risk producing unbalanced gametes as a result of unequal chromosomal segments exchange during crossing over, assuming that all the previously aborted pregnancies received identical recombinant chromosomes from their carrier mother.

This assumption was built on other studies findings on this particular inversion involving chromosome 8, confirming that both male and female carriers risk producing an unbalanced progeny. One of these studies was carried out by Asoka. Pal and colleagues in 2018, in which confirmed that 5 cases out of 172 couples with recurrent miscarriages were carriers of pericentric inversion on chromosomes 8 or 9 (5).

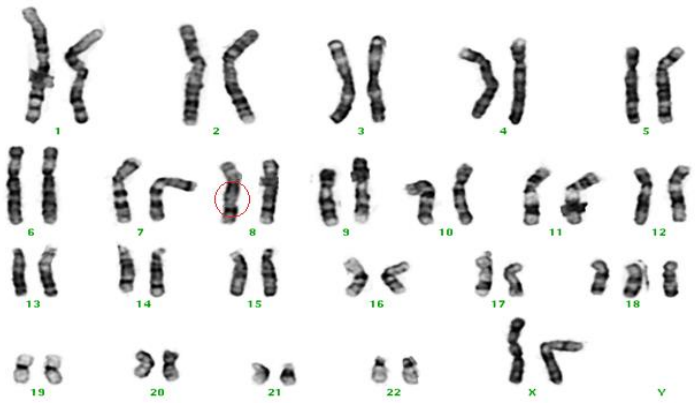

Figure1. Female karyotype shows an inversion involving both arms of one copy of chromosome 8 / 46,XX inv (8) (p11;q23)

Conclusion: These results point to chromosomal abnormalities, including pericentric inversion, as a potential major source of RPLs. This finding underlies the importance of cytogenetic investigations in the routine management of RPL to help affected families pursue the most appropriate advice and support. On the other hand, using this type of investigation as a regular management protocol will enable medical geneticists and obstetricians to provide accurate diagnoses and avoid needless medical intervention.

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References:

1. Stephenson MD. Frequency of factors associated with habitual abortion in 197 couples. Fertil Steril. 1996 Jul;66(1):24-9. PMID: 8752606.
2. Pillarisetty LS, Mahdy H. Recurrent Pregnancy Loss. [Updated 2023 Aug 28]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2023 Jan. Available from: https://www.ncbi.nlm.nih.gov/books/NBK554460/
3. Hanif MI, Khan A, Arif A, Shoeb E. Cytogenetic investigation of couples with recurrent spontaneous miscarriages. Pak J Med Sci. 2019 Sep-Oct;35(5):1422-1427. doi: 10.12669/pjms.35.5.678. PMID: 31489019; PMCID: PMC6717443.
4. Merrion K, Maisenbacher M. Pericentric inversion (Inv) 9 variant-reproductive risk factor or benign finding? J Assist Reprod Genet. 2019 Dec;36(12):2557-2561. doi: 10.1007/s10815-019-01601-y. Epub 2019 Nov 16. PMID: 31734857; PMCID: PMC6910900.
5. Pal AK, Ambulkar PS, Waghmare JE, Wankhede V, Shende MR, Tarnekar AM. Chromosomal Aberrations in Couples with Pregnancy Loss: A Retrospective Study. J Hum Reprod Sci. 2018 Jul-Sep;11(3):247-253. doi: 10.4103/jhrs.JHRS_124_17. PMID: 30568354; PMCID: PMC6262666.