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A rare karyotype variant causing repeated Abortions

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ABSTRACT

The purpose of this case report is to bring to light a rare chromosomal anomaly 46, XX, 9qh+ pattern in all 20 metaphases analysed by GTG banding linked to a series of repeated abortions (5), in very First Trimester of an expectant female of 29 years of age. Although 9qh+ is a normal variant, it has been observed to be associated with repeated abortions[1].

Keywords:- Chromosomal anomaly, repeated abortions, first trimester

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INTRODUCTION

Abortion is the termination of pregnancy, spontaneous or induced, before the period of viability, which is now accepted as 20 weeks of gestation or a birth weight of 500 grams.

Of spontaneous abortions, no cause either local or systemic, can be found in more than half. Many cases are due to faulty development of the ovum, for which genetic factors are to blame.

Nearly half of the abortuses which are chromosomally abnormal are autosomal trisomies, approximately 30% monosomies, 13% triploids, and the remainder made up of mosaics, double trisomic and tetraploid abortuses[2].

MATERIALS AD METHODS

A. *Biochemical Analysis*:-
Sample : Blood, Urine
Methods: ELISA, Chemo luminescence, HPLC.

B. *Ultrasonographical Analysis*:
Ultrasound of Gravid Uterus, Abdomen, Pelvis.

C. Early Antenatal Scan
D. *Chromosomal analysis:* sample: Blood. Lymphocyte Cultures

Biochemical report of glycosylated Hemoglobin indicated glycemic control.

Laboratory report for haemoglobin was found to be 10.5 gm% with blood group and Rh factor – B positive
ELISA for Anti-phospholipid IgG and IgM antibodies was Negative.

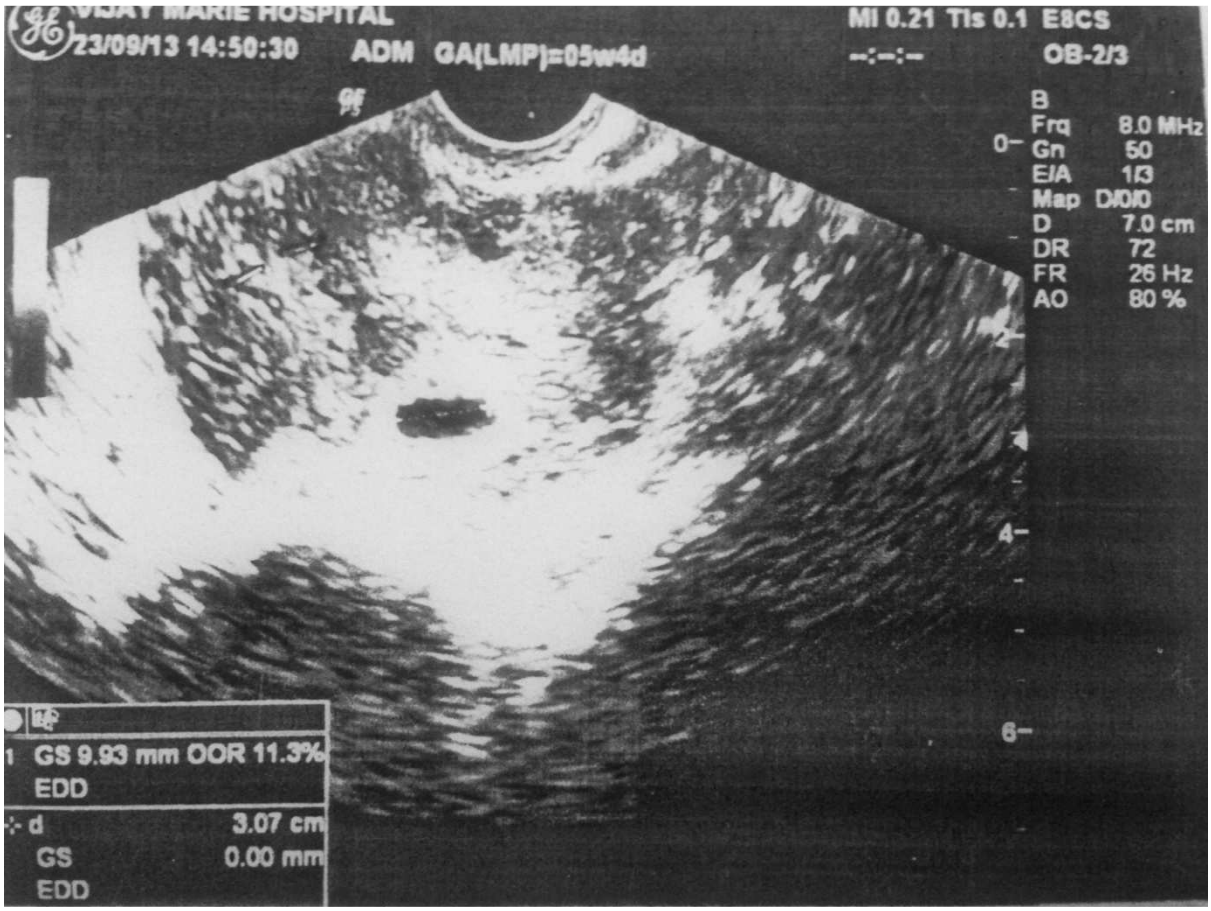

Fig.1 Ultrasound of pelvis

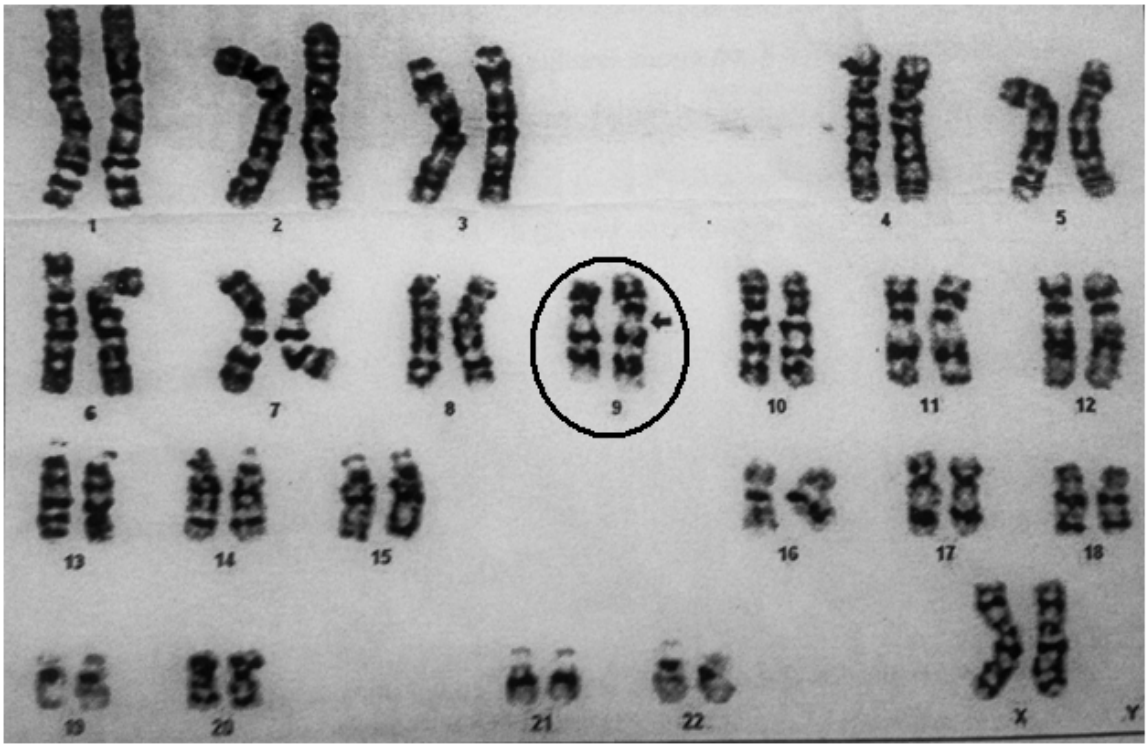

Fig.2 Karyogram of metaphase showing a female karyotype (46,XX,9qh+)

Ultrasound of Gravid Uterus, Abdomen, Pelvis showed no obvious congenital anomalies at that gestational stage; normal liver, gallbladder, spleen, pancreas, kidneys, urinary bladder.

Early antenatal scan showed signs of missed abortion of 6 weeks 23 days.

Chromosomal analysis showed a normal variant in chromosome No. 9.

RESULTS

On Chromosomal analysis of lymphocytes revealed a normal karyotype 46,XX,9qh+ pattern in all the studied 20 metaphases by GTG Banding. Further- more the Karyogram of an expectant pregnant woman showed an increase in length of heterochromatin resulting in a series of repeated abortions in the very first trimester.

DISCUSSION

Dr.Priya Selvaraj 2009 (MD MNAMS, MCE) says that the most commonly encountered polymorphism are lengthening (or) shortening of the heterochromatin of the long arm of chromosomes which are designed as 1,9,16, 9qh+. In this case, a female karyotype with an increase in length of heterochromatin on long arm of chromosome 9 has been noted, which is said to be a normal variant. However, it has been observed to be associated with repeated abortions (5 repeated abortions).

Adam H.Balen, Howard S. Jacobs, says that Gene therapy represents the ultimate i.e., correction of the gene defect itself [2]. Genetic Therapy is the insertion of a functioning gene in to the cells of a patient to correct genetic abnormality/chromosomal anomaly [3].

The scope of altering the chromosome is still under trial in the United States for treatment of diseases from cancer to viruses, untreatable genetic disorders, Sickle cell anemia, Down's syndrome. The functioning gene was named "Crispr" by the University of Massachusetts Medical School [4].

CONCLUSION

Though Karyotype 46,XX,9qh+, so discovered in connection with our case under discussion, is a normal variant, it has been linked with the occurrence of repeated abortions in the Expectant Female of First Trimester[5]. Furthermore, the Karyogram of a pregnant woman has showed an increase in the length of heterochromatin, with the consequences of a series of Abortions in her First Trimester. The cause determined to be genetic in its etiology, calls for cure in direction of gene-editing, which shall set right the problem at the gross root level, the chromosome[6].

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