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Short Communication

Homozygosity of Pericentric Inversion of Chromosome 9 and Its Possible Role in Infertility

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Abstract

The finding of homozygosity for a pericentric inversion of chromosome 9 [inv (9)] is rare in human population. We present a case (couple) where blood was received for karyotyping with indication of history of infertility. Results revealed that husband age of 33 year was homozygous for pericentric inversion 9, with karyotype 46,XY,inv (9) (p11-q13) X2, while wife age 30 year has a normal karyotype of 46, XX.

Keywords: Homozygosity; Pericentric Inversion; Chromosome# 9; Infertility

Introduction

It is well established that pericentric inversion of chromosome 9 are among most frequent chromosomal rearrangement in human. However, the finding of homozygosity for inversion 9 is rare. Recently, Stipoljev., *et al*. [1], have published cases of homozygous pericentric inversion 9, clearly indicated that so far there were only 11 cases reported between 1997-2013. We report 12th case, where homozygos-ity of inversion 9 in man with infertility problems. Heterozygosity, chromosome #9 is such common occurrence that majority of cyto geneticists would considered them as normal variants. The incidence said to be varied 1 to 2% in the general population, despite being categorized as a minor chromosomal rearrangement which does not express in phenotypes. In our retrospective study of 4500 couples referred to S. N. Gene Lab and Research Centre between 2004 and 2013 for chromosomal analysis we found only one case of homozygosity of pericentric inversion of chromosome 9 with infertility problem. A couple was referred for investigation of primary infertility.

Results and Discussion

Cytogenetic analysis was performed with GTG banding technique and karyotype was prepared and analyzed by automatic karyotyp-ing software. The wife was found to be normal having 46, XX karyotype, while husband was having homozygosity of chromosome 9 that is 46 XY, inv (9) (p11-q13) X2 karyotype (Figure 1). Figure 2(A) indicates partial karyogram with normal chromosome #9, while Figure 2 (B) shows homozygous pericentric inversion of chromosome #9. The cytogenetic analysis of parents of couple could not be carried out due to non-availability of blood.

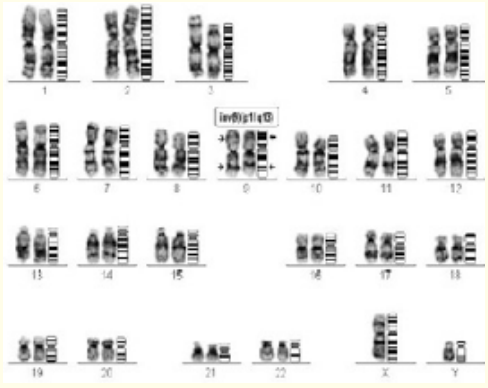

Figure 1: Karyotype of pericentric inversion on the both chromosome 9 (46, XY. Inv 9 p11 q13) X 2.

It is known that inv (9) homozygosity results from the high frequency of inv (9) heterozygosity. There are few reports indicate published cases of homozygosity with pericentric inversion 9 [2-4]. Recent report by stipoljev., et al. [1] has further shown that expected frequency of inversion 9 homozygotes in general population is 1: 40,000 calculating from the estimated frequency of heterozygotes of average 1% in general population. Rarity of the homozygosity for inv

(9) is difficult to analyze but higher prevalence of infertility has been reported in heterozygous pericentric inversion 9 of either male or female [2]. There is a need to study each breakpoint region of inv (9) using molecular cytogenetics especially for infertility using SNP array for its clinical significance.

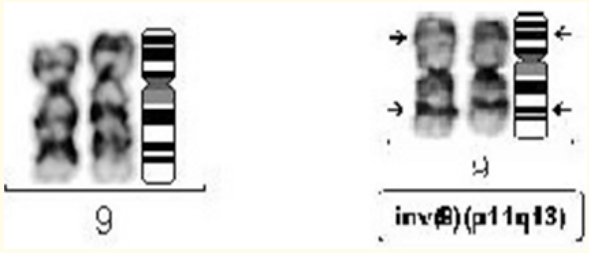

Figure 2: Parrial karyogram showing (A) normal chromosome 9, (B) Homozygosity of chromosome 9.

Conflict of Interest

Authors declare no conflict of interest.

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Volume 1 Issue 1 June 2017