Clinical significant of pericentric inversion of chromosome 9: A case report of recurrent abortion

Arefi S. (M.D.)¹, Moddaressi M.H.(M.D., Ph.D.)(2,3), Jeddi Tehrani M.(Ph.D.)⁴, Azizi F.(M.Sc.)⁵.
1-Assistant professor, Department of Reproductive Endocrinology & Embryology, Avesina Research Center, Tehran, Iran.
2-Assistant professor, Department of Genetic, Faculty of Medicine, Tehran University of Medical Science, Tehran, Iran.
3-Assistant Professor, Department of Reproductive Genetic & Biotechnology, Avesina Research Center, Tehran, Iran.
4-Assistant professor, Department of Reproductive Immunology, Avesina Research Center, Tehran, Iran.
5-Instructor, Department of Reproductive Genetic & Biotechnology, Avesina Research Center, Tehran, Iran.

Abstract

Pericentric inversion of chromosome 9 is a common phenomenon which is considered as normal variant by some cytogeneticists. A phenotypically normal couple was referred for cytogenetic evaluation due to ten recurrent spontaneous abortions and one intrauterine fetal death. The history of infertility, recurrent abortion and intrauterine fetal death were seen in the family and in the first and second degree relatives of the female. Chromosomal analysis from peripheral blood was performed according to standard cytogenetic methods using G-banding and C-banding techniques. Husband’s karyotype was normal. Wife’s karyotype has shown pericentric inversion of chromosome 9, inv(9)(p11-q13). Culture of peripheral blood, umbilical cord, chorionic villi and muscle biopsy were done on the last aborted fetus. Chromosomal study of the fetus revealed 46XY without any significant problem except for elongation of centromeric region of chromosome 9. It seems that pericentric inversion of chromosome 9 is frequently observed in individuals with recurrent abortion, intrauterine fetal death, stillbirth, but as the etiology, the clinical significance is still uncertain and we discuss this issue in our report.

Key words: Recurrent Abortion, Inversion of Chromosome 9, Intrauterine Fetal Death (IUFD), and Infertility.

Corresponding address: Dr. Arefi S., Reproductive Endocrinology & Embryology Dep., Avesina Research Center, Evin, P.O. Box 19835-177, Tehran, Iran.
Email: arefi@avesina.ir