

Chromosomal abnormalities in women with Premature Ovarian Failure

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Abstract

Introduction: Premature ovarian failure (POF) or premature menopause is one of the stressful problems in women younger than 40, which may cause numerous early or late psychological and physical complications. Failure of germ cell development is associated with complete ovarian failure, while their decreased number is more likely associated with partial ovarian failure, which leads to secondary amenorrhea. Literature review shows almost no study has been done on chromosomal factors involved in POF in Iran. The purpose of this study was to study chromosomal abnormalities in women with premature ovarian failure referred to the Fertility Health Research Center at Taleghani Hospital.

Materials & Methods: Thirty-four patients experiencing menopause before the age of 40 and resultant infertility, attended the gynecology and obstetrics ward of Taleghani Hospital of Shahid Beheshti University of Medical Sciences, from the spring of 2005 to the summer of 2006, and underwent physical exam, sonography and hormonal tests (FSH, LH and PRL). The FSH level of the cases was ≥ 40 IU/lit. Then, the patients were referred to a genetic counselor for undergoing cytogenetic tests and counseling.

Results: Most of the cases (81.8%) were younger than 40 years of age and 18.2% were 40 or older. Definite abnormal X chromosomes – mosaic karyotypes 45, X/46, XX or 47, XXY (Klinefelter's syndrome) and mosaic 46, XX or 47, XXX – were diagnosed in 17.6% of the patients (6 cases) with a mean age of 32.1 ± 7.6 .

Conclusion: Although, there were no significant correlations between POF and chromosomal abnormalities, the results from the chromosomal analysis in this study and in a limited number of cases who had been affected by early menopause due to premature ovarian failure, it is suggested that chromosomal studies be considered as one of the first steps to understand the reasons for amenorrhea. The information obtained from cytogenetic tests such as chromosomal analysis could be useful for patient management, genetic counseling, and future family planning.

Key Words: Premature ovarian failure (POF), Secondary amenorrhea, Chromosomal anomalies, Karyotype, Mosaic, Mutation.

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