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Sebnem Alanya Tosun[1], Alptekin Tosun[2]* and Enis Ozkaya[3]

[1]Department of Obstetrics & Gynecology, Bulancak Hospital, Giresun
[2]Department of Radiology, Giresun University Hospital, Giresun
[3]Department of Obstetrics & Gynecology, Zeynep Kamil Hospital, Istanbul

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***Corresponding author:** Alptekin Tosun, Department of Radiology, Giresun University Hospital, Giresun, E-mail: tosun_alptekin@yahoo.com

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Case Report

Premature Ovarian Failure in a Patient with Robertsonian Translocation Rob (14;15): Is it only a Coincidence?

Abstract

Women have fixed ovarian follicles after birth and the number of the follicles declines with age. The decrease can be regulated by genetic, hormonal and/or therapy procedures. Radiation exposure can lead to premature ovarian failure (POF). POF is defined as interruption of the ovarian function in an adolescent woman. Genetic disorders including translocations and damage on the ovarian tissue may result POF. The essential diagnostic criteria is an adolescent woman younger than 40 years of age. The diagnosis depends on at least 4 months of amenorrhea and increased FSH levels (≥40 mIU/ml) in 2 blood samples at an interval of 1 month.

Introduction

Premature ovarian failure (POF) is defined as interruption of the ovarian function in an adolescent woman before 40 years of age. Functional failure in the normal ovary leads to disturbance in the blood hormone levels which in turn leads to oligo- or amenorrhea. Low estrogen levels and increased gonadotrophins result as primary or secondary amenorrhea, and patients experience recurrence abortions [1]. The essential diagnostic criteria is an adolescent woman younger than 40 years of age. The diagnosis depends on at least 4 months of amenorrhea and increased FSH levels (≥40 mIU/ml) in 2 blood samples over a 1 month period [2]. The histopathology of the ovarian tissue obtained by mini-laparoscopic surgery can confirm the diagnosis. Bilaterally full thickness ovarian biopsy differentiates POF from resistant ovary syndrome [3].

Genetic disorders and damage on the ovarian tissue by surgery, radio- or chemotherapy may result POF. Rare causes including galactosaemia will usually diagnosed in early ages. The analysis of the family history will show variable prevalence of familiar POF up to 31%. [1,4]. X monosomy as in Turner's syndrome, X deletions and translocations may cause POF. The mutations on the phosphomannomutase 2 gene, the FSH receptor gene, the galactose-1-phosphate uridyltransferase gene, the blepharophimosis gene with chromosome 3q and the autoimmune regulator gene are the autosomal abnormalities seen in POF [1].

Case Report

A 28-year-old single nulliparous woman presented with a history of oligomenorrhea. Patient had a history of oral contraceptive use which was of inadequate dosage. Physical examination was in normal limits. Body measures were as follows: waist: 86 cm, buttocks: 100 cm. Phenotypic appearance was obese. She has 85 kg weight and 160 cm height. Her body-mass-index (BMI) was increased (33.2). Analysis of her hormone levels revealed high FSH and low E2 levels. Anti-TPO and anti-TG levels were in normal limits. In the lights of these findings, she was diagnosed as having POF. Hormone replacement therapy was prescribed to the patient. Peripheral blood sample received to research chromosomal abnormality. 20 metaphase period were analyzed with Giemsa-trypsin (GTG) banding method (300-400 bandwidth) and high resolution banding (HRB) method (550-850 bandwidth). Chromosomal analysis revealed an abnormality with 45, XX, rob (14,15) (q10; q10) chromosomal sequence. Karyotype displayed 45 chromosomes. Chromosomes of 14 and 15 were not existing to the sequence. Instead of this condition, an additional strange chromosome were obtained in the karyotype. The banding pattern of the short and long arms of the strange chromosome was showing similarity to the chromosome 14 and 15. The findings were shining lights on the presence of a nonhomologous Robertsonian translocation (RT); therefore 45, XX, rob (14,15) (q10:q10) was diagnosed.

Discussion

The average age of menarche has decreased in the recent years. Despite of decreased age of menarche, the mean age of menopause shows no change. The mean age of menopause is around the 50 years. The loss of functional follicles determines under the age of 40 years is defined as having primary ovarian failure (POF) [5]. The incidence of POF in the woman population is approximately 1-2% [6]. Most common presenting symptom is amenorrhea. Decreased levels of estrogen and increased levels of FSH and LH demonstrate on hormone sampling. Iatrogenic interventions such as radio- and/or chemotherapy, surgery may reduce the number of the follicles. Chromosomal abnormalities are also responsible. X chromosome is usually affected. XO Turner's syndrome, 47, XXX, X chromosome mosaics (45,XO/46,XX, 46 XX/47,XXX, 45,XO/46,XX/47,XXX), X chromosome deletions and inversions and X;autosome balanced translocations [5]. Patients with Robertsonian Translocation usually

present with history of recurrent abortions and hence POF may be seen in a teenage woman as well.

Chromosomal analysis is essential in both men and woman. The defect on the chromosomes have a significant effect on the fertilization. Chromosomal abnormalities have high incidence in the female gender than males [7]. RT may occur by mutation or separation in the chromosomes. The individual has normal phenotype appearance, hence has a potential for unbalanced gametes and offspring [8,9].

Cytogenetic analyses of teenage woman with POF has an importance to research the probable existance of chromosomal abnormalities. In cases less than 40 years of age, POF appears infrequently. After analysis of adequate genetic research, patients can get prompt diagnosis and also avoid unnecessary therapies. This approach leads to inform patient for the future reproductive decision.

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