JMM
pISSN: 2288-6478, eISSN: 2288-6761
http://dx.doi.org/10.6118/jmm.2015.21.2.112
Journal of Menopausal Medicine 2015;21:112-114

Case Report

Autosomal Translocation Patient Who Experienced Premature Menopause: A Case Report

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Premature ovarian failure (POF) is a condition in which the ovarian functions of hormone production and oocyte development become impaired before the typical age for menopause. POF and early menopause are present in a broad spectrum of gonad dysgenesis, from a complete cessation of ovarian function to an intermittent follicle maturation failure. Actually POF has been identified as a genetic entity (especially chromosome X), but data on genetic factors of premature menopause are limited. Until now, several cases revealed that inactivation of X chromosomes has an effect on ages of premature menopause and females with balanced or unbalanced X-autosome translocations can have several reproductive problems. On the other hand, there have been a few data that was caused by autosome-autosome translocation can lead. Therefore we report a relevant case of POF with translocation between chromosomes 1 and 4. She had her first menstrual period at the age of 12, and after 7 years she stopped menstruation. Chromosomal analysis showed 46, XX, t (1;4) (p22.3;q31.3). While evaluating this rare case, we could review various causes (especially genetic factors) of POF. To remind clinicians about this disease, we report a case of POF caused by autosome-autosome translocation with a literature review. **(J Menopausal Med 2015;21:112-114)**

Key Words: Autosomal translocation, Premature menopause, Premature Ovarian Failure

Introduction

Recently the mean age at menopause is over 45 years in 88% of women, under 45 years in 9.7%, and under 40 years in only 1.9%.[1] There have been some researches about premature ovarian failure (POF), hormone and chromosome.[2~4] The loss of functional follicles occurring in women under the age of 40 is defined as POF.[5~7] POF is a condition in which the ovarian functions of hormone production and oocyte development become impaired before the typical age for menopause.[8] POF and early menopause conditions are present in a broad spectrum of gonad dysgenesis, from a complete cessation of ovarian function to an intermittent follicle maturation failure.[9] And actually POF has been identified as a genetic entity (especially chromosome X), but data on genetic factors of premature menopause are limited.[10] So, there have been whether patients of premature menopause are associated with genetic factors. Until now, several cases reveal that inactivation of X chromosomes has an effect on ages of premature menopause and females with balanced or unbalanced X-autosome translocations can cause several reproductive problems, as expected.[11] On the other hand, there were some data that autosome-autosome translocation can lead to premature menopause.[5] And we report one case of premature menopause in infertile women with translocation between

Received: January 26, 2015 **Revised:** August 6, 2015 **Accepted:** August 6, 2015
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chromosomes 1 and 4.

Case Report

A 29-year-old Korean woman (148 cm, 35 kg) was admitted to our obstetrics and gynaecology hospital, and her chief complaint was amenorrhea for 9 years. She had no obstetric history and medical history. Her first menstruation occurred at the age of 12 and had a menstrual cycle of about 28 days. Menstruation lasted usually 3 to 5 day. But menstruation stopped after 7 years. Primary care clinic referred her to a tertiary referral hospital. Laboratory investigations showed prolactin level of 3.8 ng/mL and others were within the normal range (no abnormalities were found on complete blood count [CBC], liver and kidney panel, T3 1.13 ng/mL, Free T4 1.31 ng/dL, thyroid stimulating hormone [TSH] 0.97 uIU/mL, estradiol < 10 pg/mL, follicle stimulating hormone [FSH] 47.4 mIU/mL). On genotypic test, her karyotype is 46, XX, t (1; 4)(p22.3; q31.3). Balanced translocation between chromosome 1p22.3 and 4q31.3 had occurred. Bone marrow density (BMD) test showed T score of −37 that she had osteoporosis. Ultrasonography demonstrated non specific finding except that both side of adnexa were not seen.

Discussion

There is a patient who was diagnosed with a 46, XX, t (1; 4) (p22.3; q31.3) karyotype and her menstruation ceased since 19 years of age. POF occurs in about 1% to 2% or women,[12] and like this case, in some as early as their teens.[5] POF is a condition in which the ovarian functions of hormone production and oocyte development become impaired before the typical age for menopause.[8] POF and early menopause are present in a broad spectrum of gonad dysgenesis, from a complete cessation of ovarian function to an intermittent follicle maturation failure.[9] This reported patient's main reason of premature menopause is supposed to the translocations of different autosomal chromosomes, 1 and 4. Autosomal translocations are uncommon in women with POF and reports of translocations are X; autosome balanced translocations, with no common autosomal breakpoint.[5] As emphasized in the introduction, most genetic causes of POF were involved in X chromosome, but autosomal chromosomes can affect premature menopause and POF as well.[13] Very few cases were reported that occur translocation between autosomal chromosomes, and we can find some POF cases of 46, XX, t (2; 11), 45, XX, t (13; 14) and 46, XX, t (2; 15).[5,14] So, it can be inferred that autosomal chromosomes also work POF and Premature menopause. Especially, several reports discover that luteinizing hormone receptor (LHR; 2p21), FSH receptor (FSHR; 2p21), inhibin-alpha (INHA; 2p33-q36), forkhead box L2 (FOXL2; 3q23), estrogen receptor-alpha (ERα; 6q25), splicing factor 1 (SF1; 11q13), estrogen receptor-beta (ERβ; 14q23.2) and cytochrome P450, family 19, subfamily A, polypeptide 1 (CYP19A1; 15q21.1) of autosomes can cause POF.[13,15~18] Therefore, we can guess that autosomal chromosomes contain many unknown gene factors related to POF. Thus, to figure out how autosomal chromosomes affect POF and premature menopause, not only more POF case reports of autosomal chromosome translocation but also further studies about premature menopause are needed.

Acknowledgements

This work was supported by the Soonchunhyang University Research Fund and the Soonchunhyang University Nichebuster in Iatroscience, Creativity Education (NICE) Center.

Conflict of Interest

No potential conflict of interest relevant to this article was reported.

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