Normogonadotropik primer amenore nedeni; fibrodisplazi ossifikans progresiva

Normogonadotropik primary amenorrhea; fibrodysplasia ossificans progressiva

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Özet

Amaç: Fibrodisplazi ossifikans progresiva (FOP) ilerleyici heterotropik ossifikasyonla birlikte konjenital kemik anomalilerinin görüldüğü nadir bir hastalıktır. Yenidoğan zamanında büyük dil, el ve ayak başparmaklarında mikrodaktili, süt çocuğu döneminde aşı yerlerinde aşırı reaksiyon ve daha çok boyun ve sırt bölgelerinde başlamak üzere ağrılı kemik oluşumlarıyla kendini gösterir. (FOP)'un kadın reprodüktif sistemine etkisi tam olarak anlaşılamamıştır. Literatürde bu hastalıkla beraber canlı doğum yapmış 2 ve istemli tahliye olmuş 1 olgu mevcuttur. FOP, belki de literatürde vaka sayısı arttıkça primer amenoreye yol açan nedenler arasında yerini alacaktır.

Vaka: 18 yaşında virgin, kadın hasta , (FOP) tanısı ile adet görememe şikayeti ile hastanemize başvurdu. Hastanın sekonder seks karakterleri normal gelişmesine rağmen menstrüel siklüsü henüz başlamamıştı. Yapılan vajinal muayenede vajinal obstrüksüyonla uyumlu olabilecek anatomik bozukluk ve ultrasonografide hematokolposla uyumlu olabilecek görünüm saptanmadı. Ardışık olarak yapılan 6 aylık siklik tedavinin devamında da menstrüel siklüs başlatılamadı. Bunun üzerine hastaya endometrial biopsi önerildi. Endometrial biopsi hasta tarafından kabul edilmediği için yapılamadı.

Tartışma: (FOP) oldukça nadir görülen, kadın üreme sistemini ne şekilde etkileyeceği bilinmeyen bir hastalıktır. Her ne kadar literatürde bu hastalığa sahip üç ka-

Abstract

Objectives: Fibrodysplasia ossificans progressiva (FOP) is a severely disabling heritable disorder of connective tissue characterized by congenital malformations of the great toes and progressive heterotopic ossification that forms qualitatively normal bone in characteristic extraskeletal sites. In the literature, two women with FOP had live birth an one woman had voluntery abortion. In our case, we report an 18 years old girl with FOP who suffered from primer amenorrhea. The affect of FOP to the women reproductive system has not been understood yet by time maybe FOP may take place between the reasons of primer amenorrhea.

Case: An 18 years old, virgin patient who had the diagnosis of FOP applied to our hospital suffering from amenorrhea. Although the normal development of sekonder sexuel development she had no menstruel bleeding. With the 10 days of intervals, totally 6 hormonal measurements were performed and the results were in normal range. Neither pelvic examination nor pelvic ultrasonography determined pelvic-vaginal abnormality such as vaginal obstruction or hematocolpos. Theoretically uterus should not be affected by FOP. In our case, even the consecutive cyclic treatment for 6-month the menstrual bleeding could not be started. This situation may be explained by the unknown mechanisms of FOP which can influence the menstrual cycle.

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dında gebelik saptanmış olduğu bildirilse de, kadın reprodüktif sistemi ve (FOP) arasındaki ilişki net değildir. Bizim olgumuzda verilen 6 aylık siklik tedaviye rağmen menstrüel siklus dahi başlatılamamıştır. (FOP)'lu olguların azlığı ve mevcut olan iskelet sistemi anomalileri nedeniyle farklı sistemlere yoğunlaşılması bu hastalığın reprodüktif sisteme etkileri açısından değerlendirilmesini çok sınırlı bırakmıştır. Literatürde olgu sayısı arttıkça reprodüktif sisteme etkileri daha iyi anlaşılacak belki de (FOP) normogonadotropik primer amenore nedenleri arasında yer alabilecektir.

Anahtar kelimeler: Miyositis ossifikans; amenore; menstrüel siklüs

Discussion: As the number of cases increase in the literature, the effects of FOP to the the reproductive system will be better understanding, perhaps FOP may take place among the causes of normogonadotropik primary amenorrhea.

Key words: Myositis ossificans; amenorrhea; menstrual cycle

Introductıon

Fibrodysplasia ossificans progressiva (FOP) is a severely disabling heritable disorder of connective tissue characterized by congenital malformations of the great toes and progressive heterotopic ossification that forms qualitatively normal bone in characteristic extraskeletal sites[1-3]. The worldwide prevalence is approximately 1/2,000,000[1-3.] Most cases are sporadic and only 2 instances of familial transmission, have been documented, suggesting an autosomal dominant mode of inheritance with possible somatic mosaicism[4]. There is no ethnic, racial, gender, or geographic predilection to FOP[4-6]. FOP is characterized usually beginning in the first decade of life. The affect of FOP to the women reproductive system has not been understood yet. In the literature, two women with FOP had live birth[7-8] an one woman had voluntery abortion[9]. In our case, we report an 18 years old girl with FOP who suffered from primer amenorrhea.

Case report

An 18 years old, virgin patient who had the diagnosis of FOP 10 years ago applied to our hospital suffering from amenorrhea. She had serious scollosis and microdactyly of thumb. Sexual maturation was measured using Tanner staging[10] based on secondary sexual characteristics including the development of breasts and pubic hair. She was at Tanner stage 4 for breast and Tanner stage 5 for pubic hair. Although the normal development of sekonder sexuel development she had no menstruel bleeding. According to the genetical analysis; she had 46 XX chromosomal structure. With the 10 days of intervals, totally 6 hormonal measurements were performed and the results were within the range of 5-18 for follicle stimulating hormone, 4-16 for luteinizing hormone, 34-674 for estradiol. Thyroid stimulating hormone, prolactin, testosterone, 17 hydroxyprogesterone values were normal. Neither pelvic examination nor pelvic ultrasonography determined pelvic-vaginal abnormality such as vaginal obstruction or hematocolpos. Uterine and ovarian sizes and images on ultrasound were normal. In our case, combined estrogen-progesterone therapy was used for 21 days to start the menstrual cycle. The treatment was paused for 7 days. Consecutive cyclic treatment for 6-month was administrated but menstrual cycle could not be started. Endometrial biopsy was adviced but it was refused by the patient.

Conclusıon

The primary amenorrhea cases with normal secondary sexual development is divided to 2 groups, according to their anatomical anomalies. Mullerian anomalies, androgen insensitivity, real hermaphrodites, Asherman's syndrome can be the reasons of anatomic causes of amenorrhea. Chromosomal anomalies, radiation-chemotherapy history, infections, autoimmune disorders, and diseases such as savage syndrome occur the non-anotomical reasons of primary amenorrhea[12]. FOP a very rare disease, in what way will affect the female reproductive system is unknown. Although 3 pregnancy were reported in the literature, the relationship between FOP and female reproductive system is not clear[8-11]. Uterus have smooth muscles. Cardiac muscle and smooth muscle are not involved in the FOP process[1-5]. Theoretically uterus should not be affected by FOP. In our case, even the consecutive cyclic treatment for 6-month the menstrual bleeding could not be started. This situation may be explained by the unknown mecha-

nisms of FOP which can influence the menstrual cycle. Very limited numbers of cases and focusing skeletal abnormalities on this disease, cause the affects of FOP to the female reproductive system unknown. As the number of cases increase in the literature, the effects of FOP to the the reproductive system will be better understanding, perhaps FOP may take place among the causes of normogonadotropik primary amenorrhea.

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