



PRIMARY AMENORRHEA OF 16-YEAR-OLD GIRL THAT NO CHROMOSOMAL ABNORMALITIES

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Abstract- Primary amenorrhea is defined meaning of no menses by 14 years of age in the absent secondary sexual characteristics or no menses by 16 years of age in the present normal growth and secondary sexual characteristics. Primary amenorrhea, so far from being disease, is a symptom that results from several different causes, including developmental abnormal the genital organs, chromosomal abnormality, non-functioning of ovaries and delayed pubertal development. Also, depending on the cause of primary amenorrhea, diagnosis and adequate management as soon as possible is required, this can occur to prevent or minimize the problems.

In this case, 16-year-old woman was diagnosed as primary amenorrhea. She had no menarche and was absence of secondary sexual characteristics. She was performed chromosome analysis, hormonal level evaluation, and ultrasonography (USG). She was diagnosed primary amenorrhea as pituitary originated hypogonadotropic hypogonadism, and estradiol hormonal treatment was prescribed.

Keywords- Primary amenorrhea, Secondary sexual characteristics, Sexual hormone

Introduction

Primary amenorrhea is defined as delayed menarche by 14 years of age in the absence of secondary sexual characteristics or absence of menses by 16 years of age in the presence of normal growth and secondary sexual characteristics [1]. Primary amenorrhea, so far from being disease, is a symptom that results from several different causes, including developmental abnormal the genital organs, chromosomal abnormality, non-functioning of ovaries and delayed pubertal development [2,3]. The most common causes are hypothalamic amenorrhea, polycystic ovarian syndrome, hyperprolactinemia, and ovarian failure [1]. The patient with primary amenorrhea depending on its causes, therefore, early diagnosis and appropriate management are necessary in order to prevent or minimize the problems [4].

Case Report

A 16-year-old girl did not have menstruation that visited the hospital outpatient clinic. First, check the history of the patient and performed the physical and gynecologic examination particular attention. She had no pubarche, no axillary hair and no breast budding. Eventually, she was absence of secondary sexual characteristics. In past medical history, she was treated with chemotherapy for childhood hemangioma. However, failing to remember the exact chemotherapeutic agents were. She was performed chromosome analysis, estradiol (E2), follicle-stimulating hormone (FSH), luteinizing hormone (LH), prolactin, complete blood cell count, serum chemistry, and ultrasonography (USG). Chromosome study showed normal female karyotype and laboratory test result was as following; E2 < 10 pg/ml, FSH 0.2 mIU/mL, LH 0.04 mIU/mL, prolactin 7.6 ng/mL, and glucose 129mg/dL. On USG image finding showed very small uterus, 1.2 mm in endometrial thickness [Fig-1]. But, both ovaries on USG did not show. The patient was diagnosed as pituitary originated ovarian dysfunction, and estradiol hormonal treatment was prescribed.



Fig. 1- USG finding is sagittal view of 1 cm sized small uterus (↑).
UT: Uterus, BL: Bladder

Discussion

The practice committee of the American Society for Reproductive Medicine (ASRM) suggests that, owing to a trend to earlier age at menarche, evaluation of primary amenorrhea should take place by the age of 15 for adolescent females presenting with normally developed secondary sexual characteristics [5]. Moreover, adolescent girls who have not reached menarche within 5 years after thelarche and amenorrheic girls who present without secondary sexual characteristics development until the age of 13, should also begin evaluation [6]. Primary amenorrheic patient first in case of examinations is a closed medical history, and should contain data for family history, any systemic disease of the amenorrheic patient. In patient with primary amenorrhea, the presence of developed secondary sexual characteristics is important and demonstrates that sexual steroids are formed and circulating [5]. During physical examination carefully, it should be searching for signs or symptoms of any underlying systemic disease and the presence of developed secondary sexual

characteristics. Gynecologic examination per rectum bimanual evaluation can verify anatomical existent or absent of the uterus and palpable masses at the adnexa, but in recent years, such as ultrasonography (USG) or computed tomography (CT) radiological methods have changed. Laboratory tests and radiography, if indicated, should be performed to evaluate for suspected systemic disease [7]. During performed the physical examination, she had no pubarche, no axillary hair and no breast budding. USG finding showed small uterus and not seen both ovary.

Young women with primary amenorrhea and absent secondary sexual characteristics development should start first laboratory evaluation with check with follicle-stimulating hormone (FSH) and luteinizing hormone (LH). Elevated gonadotrophin levels imply that the cause of the delayed puberty lies within the gonads, while low or normal FSH and LH levels point to constitutional delay of puberty, pituitary dysfunction, or hypothalamic disorders [5]. Since secondary sexual characteristics were absent in amenorrheic patient, the diagnosis relied on laboratory test results and karyotype analysis. Primary amenorrhea is often associated with disorders of endocrine function, with gonadal and somatic anomalies but it is an uncommon presentation in reproductive medicine [8]. In our patients, chromosomal study was normal karyotype, was performed laboratory tests. The patient turned out to be having hypogonadotropic hypogonadism, with low FSH level (0.2mIU/mL) and low LH level (0.4mIU/mL). She also showed low estradiol level below 10pg/ml. Chronic illnesses like chronic liver disease, chronic renal insufficiency, diabetes, immunodeficiency, inflammatory bowel disease, thyroid disease, severe depression or psychosocial stressors can also cause hypogonadotropic hypogonadism [1].

Reproductive medicine has changed dramatically since the 1981 publication of the study of patients presenting with pubertal amenorrhea [9]. The breakdown of causes likely remains unchanged, with the four most common causes of primary amenorrhea being ovarian failure (48.5%), congenital absence of the uterus and vagina (16.2%), gonadotropin-releasing hormone (GnRH) deficiency (8.3%) and constitutional delay of puberty (6.0%) [10]. Other causes include Excessive exercise, excessive weight loss or malnutrition, and anorexia or bulimia nervosa. In addition, possibility of prior cranial radiation or systemic chemotherapy or presence of central nervous system tumor cannot be excluded [5]. Our patient was treated with chemotherapy for childhood hemangioma. The exact chemotherapeutic agents do not really know, but alkylating agents is probably assumed. Pituitary originated hypogonadotropic hypogonadism caused by these agents is thought.

Treatment is that hormone therapy, estrogen replacement therapy is needed because hypogonadotropic hypogonadism.

If a patient has no secondary sexual characteristics and no menarche like this case, primary amenorrhea can be diagnosed as early as 14 years of age [4]. If not treated, however, the patient can lead to serious health risks and problem over a long term including estrogen deficiency resulting problems, sexual dysfunction, infertility, and innate problems of the underlying health risks. Also, depending on the cause of primary amenorrhea, diagnosis and adequate management as soon as possible is required, this can occur to prevent or minimize the problems.

Though the patient is having hypogonadotropic hypogonadism, more detailed cause in this patient remains unknown for now. It can be either hypothalamus or pituitary dysfunction, and stimulation test by intravenous gonadotropin-releasing hormone (GnRH) agonist

can help distinguishing the origin of amenorrhea. Additionally, other causes than direct disorder in hormone production should be investigated and radiologic methods like magnetic resonance imaging (MRI) should be performed.

Conflicts of Interest: None declared.

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