Batı Karadeniz Tıp Dergisi
Medical Journal of Western Black Sea

Doi: 10.29058/mjwbs.2018.2.5

Olgu Sunumu

Mayer-Rokitansky-Küster-Hauser Sendromu: Atipik Bir Şikayet ile Başvuru

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M A K A L E
B İ L G İ S İ

Gönderilme Tarihi:
20.12.2016
Revizyon:
25.06.2018
Kabul:
18.07.2018

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Anahtar Kelimeler:
Konjenital anomali, primer amenore, inkontinans

Ö Z E T

Mayer-Rokitansky-Küster-Hauser (MRKH) sendromu, normal sekonder seks karakter ve dış genital gelişimi ile birlikte uterus, serviks ve vajina yokluğu ile karakterize konjenital bir anomalidir. Dişi cinsiyette 4000 ile 10.000 doğumda bir görülen nadir bir sendromdur. Genellikle puberte döneminde primer amenore etiyolojisinin araştırılması sırasında tanımlanan sendrom, çeşitli klinik şikayetlere de neden olabilir. Ancak literatürde, işeme şikayetleriyle kliniğe başvurarak teşhis edilen vaka bildirilmemiştir. Üriner inkontinans kadınlarda yaygındır, ancak yaşam kalitesini etkilese de bu durumun doğal bir süreç olduğu düşünülerek tıbbi yardım arayışına girilmez ise altta yatan önemli faktörlerin tanısında gecikme yaşanabilir. Klinik pratikte bu hastaların doğru tanınması için, herhangi bir nedenle kliniğe başvuran hastalarda inkontinans varlığı hekimler tarafından mutlaka sorgulanmalıdır.

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Case Report

Mayer-Rokitansky-Küster-Hauser Syndrome: Admission With An Atypical Chief Complaint

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ARTICLE INFORMATION

Date of Submission
20.12.2016
Revision:
25.06.2018
Accepted:
18.07.2018

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Key Words:
Congenital anomaly, primary amenorrhea, incontinence.

ABSTRACT

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a congenital abnormality with absence of uterus, cervix and vagina, together with normal sex development and external genitalia. It's a rare case of girls seen in every 4,000 to 10,000 births. The syndrome, usually identified during the investigation of primary amenorrhea in puberty, may have additional abnormalities, that also can cause a variety of clinical complaints. However, in the literature there is no patients diagnosed as a result of admission with complaints of micturition. Urinary incontinence is common among women but while they do not seek for medical help and while they think that it's a natural process though the situation affects their quality of life, may delay the diagnosis of important factors that may lie beneath. For the correct identification of these patients in clinical practice all female patients admitted to the clinic for any reason incontinence should be noted by physicians.

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Introduction

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, is a congenital aplasia of uterus and upper (2/3) vagina resulting from a pause in embryological development of the mullerian ducts [1-3]. MRKH syndrome is very rare with an incidence of 1 in 4500 births[4]. According to the clinical findings, it is divided as type 1 (isolated uterovaginal defects) and type 2 (additional renal, cardiac, vertebral and auditory anomalies) [5]. Growth and development and ovarian functions are normal with 46, XX karyotype analysis[2,3]. Secondary sexual development is also normal[1].In 1/3 of patients, urinary tract abnormalities, and in 10% of patients skeletal abnormalities may be associated with. It is the second most frequent cause of primary amenorrhea after gonadal disgenezis. It is the cause of 15-20% of all primary amenorrhea [2]. These cases of which the etiology is not fully clarified, generally come to clinic with primary amenorrhea [4]. Although there are cases in the literature diagnosed with different clinical applications, there is no patient admitted with voiding complaints. In this regard, in our case, the importance of a detailed examination in patients with voiding complaints has been emphasized.

Case

In the clinical interrogation of a 29 year-old woman that admitted to our clinic with voiding complaints, it was learned that she had polakuria, nocturia, urgency since childhood and recently increased urgency type of incontinence which was also the chief complaint of the patient. The patient with low socio-economic aspects had not seek professional help before, and had tried to overcome the problem with the help of her mother. It was learned that recently she had also complains of constipation. Urine analysis and urine culture showed no abnormal findings. There was not any abnormality at complete blood count and blood biochemistry. The Uroflowmetry performed in a clinical setting maximum urinary flow (Qmax) was 24 ml/ sec and volume voided in 24 sec was 240 ml. There was no post-voided residual urine. At urinary system ultrasonography where the left kidney was not observed, the 101x70 mm sized right kidney was observed in the right iliac fossa (ectopic kidney).

At abdominal computed tomography taken in this case with solitary ectopic kidney in order to identify possible additional pathologies, at pelvic level slightly right to the midline a solitary ectopic kidney with a long axis of 104 mm and average parenchymal thickness of 2.6 cm was observed. Uterus was not seen. At the locations of ovaries there were no pathological findings detected (Figures 1 and 2).

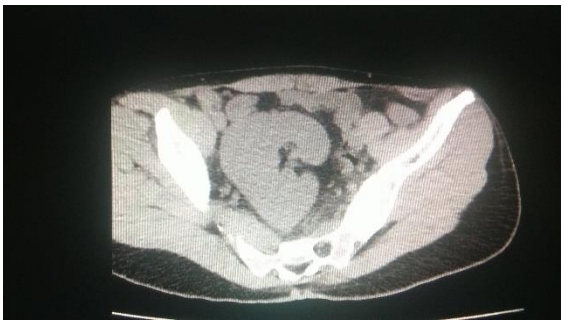

Figure 1: Solitary ectopic kidney at pelvic level 4

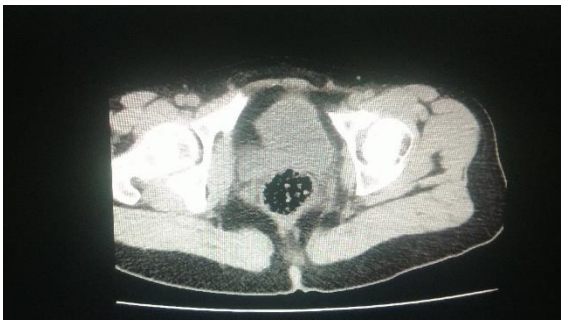

Figure 2: No uterus seen

The Tc-99m -DMSA test was reported as ‘’The right kidney is not in the normal localization, it locates in the right pelvic area. As far as can be assessed, it was in the normal size, radiotracer uptake was observed homogenously. Left kidney is not observed in normal localization, an adjacent view to the lower pole of the ectopic right kidney is available. As far as the evaluation, the size of the left kidney was small and radiotracer uptake was observed to be reduced.’’

The secondary sex character of the patient who described primary amenorrhea at clinical questioning was found to be normal. Hormone profile was normal. She had 46, XX karyotype in genetic screening and additional genetic abnormalities were not detected. It was observed that patient with natural cardiological evaluation had pectus excavatum deformity. In the light of these findings, patient was diagnosed as type 2 MRKH syndrome and lifestyle changes were suggested for the patient's voiding problem due to the mechanical pressure of the ectopic kidney after taking the patient's written consent.

Discussion

Urinary incontinence (UI) is a common problem affecting the psychological, physical, the social and economic welfare of people and their families. At the

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same time is a major cause of clinical application in urology. To have a normal continence, a firm anatomy, neural and hormonal system is required. A damage occurring in one or more of these systems will result in incontinence. Studies done in our country indicate that families use usually traditional methods to treat incontinence in childhood[6]. With the rate of %6.4, usage of professional methods among 5-12 years age group patients supports that the families were not overly disturbed [6]. Among 13-19 years age group, 76.9% of application of professional treatment, shows that more care was given in the adolescent age group by Turkish family[6]. This explains the delayed seeking of treatment of our case.

MRKH syndrome was first reported by mayer 1829 as Mullerian agenesis. Then Rokitansky and Kuster defined this syndrome as, lack of vagina and various uterin anomaly together with normal functional ovaries and external genitalia. Hauser expanded the definition by adding some renal and skeletal abnormalities[7].

MRKH syndrome is thought to be as a result of a variety of genetic and environmental factors. Also maternal gestational diabetes and teratogen exposure was blamed [3]. Although often seen as sporadic cases today polygenic / multifactorial hypothesis has gained the power switch [8]. The absence of family history, the absence of history of gestational pregnancy and teratogen exposure in her mother in our case, suggest that this case is sporadic. Any failure in the formation of müllerian duct, may cause many abnormalities at uterin and vaginal level. They can appear combined, also can be seen individually. As the gonadal ridges was the origin of ovary, ovarian pathology with uterovaginal pathology seen rarely together, while the close relationship of wolfian channel and müllerian duct explains the developments and combinations of urinary and genital tract abnormalities [9].

For clinical diagnosis, physical examination, ultrasonography (USG), magnetic resonance imaging (MRI) and laparoscopy may be used. As clinical examination for vagina and cervix was adequate, for the uterus ultrasound and MRI, laproscopy for adnexal structures was preferred [10].

In the differential diagnosis, causes of primary amenorrhea and diseases with normal secondary sex characteristics should be considered (table 1).

Table 1:Summary of differential diagnosis between MRKH syndrome and isolated vaginal atresia,WNT4 syndrome, and androgen insensitivity syndrome [3].

	MRKH/MURCS	Isolated vaginal atresia	WNT4 syndrome	Androgen insensitivity
Upper vagina	Absent	Variable	Absent	Absent
Uterus	Absent	Present	Absent	Absent
Gonads	Ovary	Ovary	Masculinized ovary	Testis
Breast development	Normal	Normal	Normal	Normal
Pubic-hair development	Normal	Normal	Normal	Spares
Hyperandrogenism	No	No	Yes	No
Karyotype	46,XX	46,XX	46,XX	46,XY

First gonadal dysgenesis should be excluded. The differential diagnosis should include congenital absence of the vagina and uterus, isolated vaginal atresia and androgen insensitivity [11]. Transverse vaginal septum and imperforate hymen are not included in diagnosis because there is a normal vagina and uterus of the patients.

These cases can be detected usually while investigating primary amenorrhea during puberty. Ectopic kidney can cause vague abdominal pain or atypical style colic pain and cause to be misdiagnosed as acute appendicitis and pelvic inflammatory disease in women. Among other signs and symptoms of an ectopic kidney; incontinence, palpable abdominal mass, frequent urinary tract infections and renovascular hypertension may be considered [12]. In this case, voiding complaints due to mass effect of kidney which was settled in the pelvic reagion was present.

Because socioculturally low female patients were not able to express themselves sufficiently, in clinical practice can lead to delay in diagnosis. In this context, admission to the clinic because of inability to engage in sexual intercourse and late diagnosed cases are available. On the other hand, there is a higher proportion of patient among women of low

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educational level than women of high-level education that reports health-related limitations and urinary incontinence as a disturbing health problem [13]. Diagnosis of MRKH syndrome because of admission to the clinic with voiding complaints as a result of disturbance of bladder dynamics because of pelvic kidney are not available in the literature.

Conclusion

Urinary incontinence in female patients is frequent but mostly unheeded, and even is a major health problem because not enough questioned by physicians. Because patients do not apply to the physicians and resume it as a natural process though it affects the quality of their life, delays the diagnosis of important reasons that may lie beneath. Although MRKH syndrome in clinical practice is not often encountered, with accurate diagnostic tools can be easily diagnosed. The presence of additional variations and the system anomalies that participate to the syndrome, can sometimes create the first complaint in the clinic. It should be stressed that for the correct identification of these patients that are more vulnerable to psychiatric trauma, in clinical practice keeping in mind to consider in this respect and all female patients admitted for any reason, incontinence should be questioned by doctors.

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