Case Report / Olgu Sunumu

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Mayer-rokitansky-kuster-hauser syndrome with presacral schwannoma presenting as a pelvic mass: A literature review and case report

Pelvik kitle olarak prezente olan presakral schwannomlu mayer-rokitansky-küster-hauser sendromu ve literatürün gözden geçirilmesi

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Abstract

A 32-year-old woman was admitted to our gynecology outpatient clinic with primary amenorrhea, a pelvic mass, and pain. Sonographic examination and magnetic resonance imaging revealed an approximately 124×103 mm heterogeneous mass. Moreover, laparotomy revealed fibrotic uterine bands with normal ovaries, tubes, and a solid retroperitoneal lesion. On the second postoperative day, the mass was removed, and the patient was discharged with full recovery. Microscopic examination of the pelvic mass confirmed the diagnosis of schwannoma. To the best of our knowledge, this is the first report on the co-occurrence of Mayer-Rokitansky-Küster-Hauser syndrome and schwannoma, without the presence of any other pathology.
Keywords: MRKH syndrome, schwannoma, pelvic mass

Öz

Pelvik kitle olarak prezente olan presakral schwannomlu Mayer-Rokitansky-Küster-Hauser sendromu (MRKHS). MRKH'li hastalar sıklıkla ekstragenital anomaliler, özellikle ürolojik ve iskelet anomalileri sergiler. Hastamız, schwannomanın eşlik ettiği ilk MRKHS olgusudur. Otuz iki yaşında kadın hasta jinekoloji polikliniğimize primer amenore, pelvik kitle ve ağrı şikayetleri ile başvurdu. Sonografik değerlendirme ve manyetik rezonans görüntülemede, yaklaşık 124×103 mm'lik heterojen bir kitle görünümü mevcuttu. Laparotomide fibrotik uterin bantlara eşlik eden normal overler, tubalar ve sert bir retroperitoneal lezyon izlendi. Kitle ekstirpe edildi ve postoperatif ikinci günde hasta şifa ile taburcu edildi. Pelvik kitlenin mikroskobik incelemesi schwannoma tanısını doğruladı. Bilgilerimize göre bu iki malformasyonun herhangi bir ek patoloji olmaksızın birlikte ortaya çıktığı ilk olgudur.
Anahtar Kelimeler: MRKH sendromu, schwannoma, pelvik kitle

Introduction

Vaginal agenesis, also known as Mullerian aplasia or Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, is characterized by congenital absence of the vagina and variable uterine development. It results from agenesis or hypoplasia of the Mullerian duct system; however, the underlying etiology remains unknown. The incidence of MRKH syndrome has been estimated to be 1 in every 4.500 female births[(1)]. Most of the cases appear to be sporadic; however, cases having a family history of this disease have also been described. The first clinical signal is generally primary amenorrhea in patients who have a normal female phenotype, a normal 46 XX karyotype, and normal and functioning ovaries with no signs of androgen excess[(2)]. Furthermore, external examination of these patients reveals completed puberty with normal secondary female sexual characteristics (pubic hair and breast development: Tanner stage 5) and normal external genitalia.

Schwannomas are rare tumors of ectodermic origin that grow from the neural sheath and are usually found in the neck and

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extremities. These tumors commonly arise from the cranial nerves as acoustic neurinomas, but they are extremely rare in the pelvis and retroperitoneal area (less than 0.5% of reported cases), unless they are associated with von Recklinghausen disease (type 1 neurofibromatosis)[3]. Schwannomas are mostly benign in nature and are more common in adult females between the age of 20 and 50 years, with a male-to-female ratio of 2:3. Preoperative biopsy examinations can be challenging, and immunohistochemistry is necessary for the correct diagnosis of a schwannoma[4]. Further, a low rate of mitosis and the absence of atypical mitotic figures and nuclear hyperpigmentation characterize a benign lesion.

To the best of our knowledge, this is the first report on the co-occurence of MRKH syndrome and schwannoma in adulthood, without any other pathology, such as genitourinary system abnormalities, skeletal anomalies, or deafness.

Case Presentation

A 32-year-old virgin female patient was admitted to our gynecology clinic with primary amenorrhea and pain. Her family history was unremarkable. Abdominal examination revealed a rigid and fixed mass extending to the level of the umbilicus, with no visceromegaly or ascites. During the pelvic examination, the external genitalia were normal, but there was vaginal agenesis (Figure 1A). In addition, breast development was normal (Tanner stage 5). Ultrasound imaging revealed a 124×103 mm heterogeneous mass, and an abdominal magnetic resonance scan revealed a 130×100 mm mass lesion located in the presacral region and compressing the bladder posteriorly. However, the patient had no complaints related to defecation or miction. Furthermore, all laboratory findings were normal (follicle stimulating hormone: 7.9 mIU/mL, luteinizing hormone: 14.5 mIU/mL, estradiol: 200 pg/mL, Ca-125: 12.2 U/mL, CA 19-9: 8.0 U/mL, CA 15-3: 7.4 U/mL, and hemoglobin: 14 g/dL).

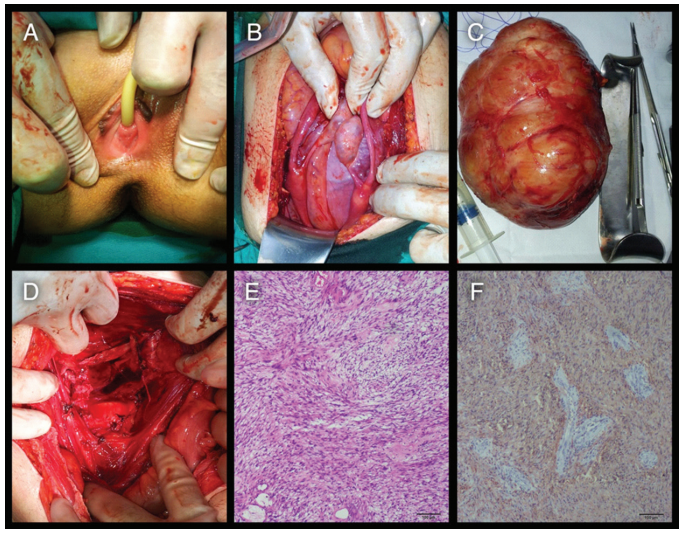

Figure 1. External genitalia (A), internal genitalia and the pelvic mass (B), appearance of specimen (C), retroperitoneal area after mass removal (D), histological appearance of schwannoma, spindle cell neoplasm composed of cellular and hypocellular areas (H&E 100x) (E), and S100 immunostaining of Schwann cells immunohistochemistry 100x (F)

The patient underwent laparatomy under general anesthesia. The tubes and ovaries were normal, but the uterus was seen as two distinct tubular-shaped fibrotic bands (Figure 1B). Behind the internal genital organs, a solid and fixed mass was discovered (Figure 1B). This mass was extirpated by a general surgeon (Figure 1C, D). There was no complication in the postoperative period and she was discharged on the second postoperative day.

A schwannoma was discovered during the pathological examination of the specimen (Figure 1E, F). Immunohistochemical markers showed a diffuse positivity of S-100, actine (-), desmin (-), Kİ-67: 3%, and CD34 (-).

The patient was asked to sign a consent form for publication.

Discussion

Women with MRKH syndrome have a normal female karyotype, with normal ovaries and ovarian functions, and thus they develop normal secondary sexual characteristics (e.g., breasts, axillary hair, and pubic hair). However, these women have a congenital absence of the uterus, cervix, and upper part of the vagina.

In the literature search we conducted using the keywords "MRKH syndrome" and "pelvic mass," very few case reports were found. In most of these cases, the pelvic mass was found to be a leiomyoma. They reported that despite the low probability of having fibroids without a uterus, fibroids should not be ruled out in such patients[5,6].

It is known that urinary system anomalies are frequently observed in patients with MRKH syndrome. Karimbayev et al.[7] presented a 14-year-old patient with MRKH syndrome and pelvic ectopic kidney as a pelvic mass in their study.

Bae et al.[8] reported the first known case of cancer of the supernumerary ovary in a patient with MRKH syndrome, and they proposed that although both ovaries were confirmed to be normal in the patient with MRKH syndrome, ovarian neoplasm should be considered in the diagnosis of a pelvic mass.

Juusela et al.[9] presented two rare pathologies, bilateral Sertoli cell tumors of the ovary and MRKH syndrome, which developed concomitantly in the same patient.

In a study carried out by Kawano et al.[10], bilateral Mullerian remnants with functioning endometrium and a pelvic mass that was considered an endometriotic cyst were revealed on magnetic resonance imaging. Table 1 shows the author's name, publication year, and the diagnosis of accompanying pelvic masses in patients with MRKH syndrome[11-20].

To the best of our knowledge, this is the first case in which MRKH syndrome and schwannoma are seen together. MRKH syndrome is mainly sporadic, but it may be an inherited disorder. The genetic defects responsible for MRKH syndrome remain unclear. Furthermore, a recent study investigated male

Table 1. Author's name, publication year, and the diagnosis of accompanying pelvic masses in patients with MRKH syndrome

Author	Publication year	Diagnosis
Papa et al.[11]	2008	Leiomyoma
Lanowska et al.[12]	2009	Leiomyoma
Fletcher et al.[13]	2012	Leiomyoma
Bae et al.[8]	2013	Cancer of the supernumerary ovary
Kawano et al.[10]	2014	Endometrioma
Girma and Woldeyes.[14]	2015	Leiomyoma
Hasegawa et al.[15]	2015	Leiomyoma
Narayanan et al.[16]	2015	Leiomyoma
Dimitriadis et al.[17]	2016	Mitotically active leiomyoma
Karimbayev et al.[7]	2018	Pelvic ectopic kidney
Juusela et al.[9]	2018	Bilateral ovarian Sertoli cell tumors
Jokimaa et al.[18]	2020	Leiomyoma
Romano et al.[19]	2021	Leiomyoma
Ibidapo-Obe et al.[20]	2021	Leiomyoma

MRKH: Mayer-Rokitansky-Küster-Hauser

microchimerism as a possible cause but found no evidence to support this finding[21]. Schwannomas may occur spontaneously or in the context of a familial tumor syndrome, such as neurofibromatosis type 1[22]. Retroperitoneal schwannomas are extremely rare tumors that are difficult to diagnose preoperatively[23].

Besides MRKH syndrome and retroperitoneal schwannoma, our patient had no other conditions. Moreover, genetic studies could not be carried out. Therefore, we do not know if there is a common genetic defect that can cause both disorders. However, since the presented case is unique in the literature, it appears unlikely that a common genetic defect exists.

Treatment for MRKH syndrome involves a combination of psychosocial support and correction of anatomical defects, such as the creation of a functional vagina and transplantation of a uterus. In our case, since the patient is not married and her primary complaint was pelvic pain, we only surgically removed the mass. She was relieved after the surgery, and since the pathological diagnosis was benign schwannoma, no further treatment was required. At a 6-month postoperative follow-up, the patient was completely asymptomatic, and computed tomography scan imaging revealed no mass.

Conclusion

We present a unique case of MRKH syndrome with a schwannoma. To treat pelvic masses with Mullerian congenital anomalies, accurate evaluation and informed consent are required prior to surgery.

Ethics

Informed Consent: The patient was asked to sign a consent form for publication

Peer-review: Externally and internally peer-reviewed.

Authorship Contributions

Surgical and Medical Practices: R.N., S.Ö., M.A.N., S.S., E.A., Concept: R.N., S.Ö., M.A.N., S.S., E.A., Design: R.N., S.Ö., M.A.N., S.S., E.A., Data Collection or Processing: R.N., S.Ö., M.A.N., S.S., E.A., Analysis or Interpretation: R.N., S.Ö., M.A.N., S.S., E.A., Literature Search: R.N., S.Ö., M.A.N., S.S., E.A., Writing: R.N., S.Ö., M.A.N., S.S., E.A.

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