An incidental coexistence of Mayer-Rokitansky-Kuster-Hauser syndrome with pelvic ectopic kidney and perirenal endometrioma

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ABSTRACT

في هذا التقرير حالة لمتلازمة ماير– روكيتانسكي– كوستير– هاوزر مع كلية منتبذة الحوض، وكيس حول الكلية، ونسيج في بطانة الرحم من الداخل. نستعرض هنا حالة مريضة تبلغ من العمر 17 عاماً تم ادخلها إلى المستشفى، تعاني من انقطاع أولي في الطمث، علماً أنها أكملت مراحل البلوغ. أظهرت نتيجة فحص الموجات فوق الصوتية عدم إمكانية اكتشاف الرحم ووجود آفة كيسية بحجم 6x11cm في منطقة التوابع اليمنى. تم اكتشاف إنتباذ حوض الكلية بقياس 5.5x9cm في الوسط. كانت الهرمونات وعلامات الورم طبيعية. أجريت الفحوصات المخبرية، وأظهرت نتيجة فحوصات تنظير البطن عدم إمكانية اكتشاف الرحم وكلا الأنبوبين، وكان المبيضين طبيعيين. كان هنالك كيس بقياس 6x7cm يقع في المنطقة خلف الصفاق. لم يتم التمكن من تحديد منشأ الكيس. أُخذ إجراء عملية فتح البطن بعين الاعتبار، أُدخل حيز خلف الصفاق وتم اكتشاف وجود كيس بريتوني طري بحجم 8x11cm مجاور لحوض الكلية، وتم استئصاله. كشفت نتائج فحص الأمراض بوجود نسيج في بطانة الرحم ونزف داخلي.

In this case report, a Mayer-Rokitansky-Kuster-Hauser syndrome with pelvic ectopic kidney and a perirenal cyst with endometrial tissue inside is demonstrated. A 17 year old patient admitted with primary amenorrhea. Pubertal stages were completed. In pelvic ultrasonography; uterus could not be detected, a 6x11 cm sized cystic lesion was seen on the right adnexal area. A centrally located 5.5x9 cm sized ectopic pelvic kidney was detected. Hormones and tumor markers were normal. Laparoscopy was planned. In the laparoscopic observation, uterus and both tubes could not be detected, ovaries were normal. There was a 6x7 cm sized cyst located in the retroperitoneal area, the origin of the cyst could not be identified. Laparatomy was considered, retroperitoneal space was entered, an 8x11 cm sized smooth contoured perirenal cyst adjacent to the pelvic kidney was detected. Cyst was extirpated. The pathology result was reported to include endometrial tissue and hemorrhage inside.

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Mayer-Rokitansky-Kuster-Hauser syndrome (MRKH) is characterized by the congenital absence of vagina and with several Mullerian duct abnormalities frequently with the aplasia of uterus. It is the second most common cause of primary amenorrhea. Its incidence is 1/4000-1/5000 live births. According to another reference is reported as 1/20.000.[1,2] Diagnosis cannot be established until the sexual intercourse takes place or the patient complains of primary amenorrhea. The mean age of diagnosis is 15-18 years. It can be frequently seen with urinary tract and skeletal system abnormalities. In this case report, an incidental coexistence of MRKH syndrome with pelvic ectopic kidney and a perirenal cyst with endometrial tissue inside is demonstrated, and is unique in literature.

Case Report. A 17 year old patient admitted to our clinic with the complaint of primary amenorrhea. In her physical examination, it was detected that the pubertal stages (pubarche and telarche) were completed. In pelvic examination vulva was normal and the depth of vagina was 8 cm. The laboratory findings were follicle-stimulating hormone (FSH): 8.7 mIU/ml (3-15 mIU/ml), luteinizing hormone (LH): 13.8 mIU/ml [2.5-9.0 mIU/ml (Mid-cycle up to 90)], estradiol: 34 pg/ml (30-49 pg/ml), prolactin: 8.77 ng/ml (4.6-37 ng/ml), dehydroepiandrosterone sulfate (DHEA-S): 208 mcg/dl (32-210 mcg/dl). The total blood count, liver, and renal function tests [creatinin: 0.8 mg/dl (0.6-1.2 mg/dl), blood urea nitrogen (BUN): 12 mg/dl (5-23 mg/dl)] were normal. The tumor markers were also normal. The chromosomal test was 46,XX. In ultrasonographic

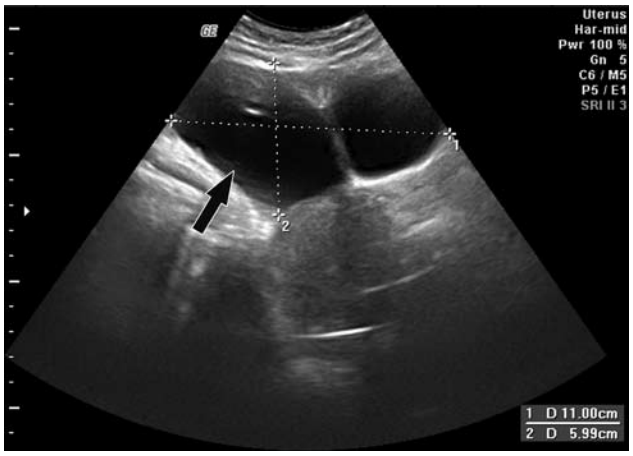

Figure 1 - The ultrasonographic image of the 6 x 11 cm sized cystic lesion on the right adnexal area (arrow).

examination, uterus could not be detected, a 6x11 cm sized cystic lesion with solid mass inside was detected on the right adnexal area (**Figure 1**). The origin of the cyst could not be clarified. In pelvic MRI, uterus could not be detected, in the pelvic region a 9x11 cm sized right sided septated cystic lesion was defined with unknown origin, and the bladder was displaced by the lesion, and a pelvic ectopic kidney was measured as 5.5x9 cm in the middle of pelvis. Laparoscopy was planned. In the laparoscopy, uterus could not be detected and both tubes and the ovaries were normal. There was a 6x7 cm sized cystic lesion thought to be located in the retroperitoneal area, and appeared on the right adnexal area, and the origin of the cyst could not be identified. Laparotomy was initiated. The laparotomic exploration was similar with laparoscopy. Retroperitoneal space was entered. Right ectopic kidney and a 8x11 cm sized smooth contoured perirenal cyst adjacent to the pelvic kidney were detected. Left kidney size and localization was normal. The cystic ingredient was aspirated and observed as hemorrhagic. Cyst was extirpated, and the frozen examination was reported as benign. Right ectopic kidney was left on its localization. The pathology result after the operation was reported to include endometrial tissue and hemorrhage inside.

Discussion. Mayer-Rokitansky-Kuster-Hauser syndrome patients usually have normally functioning ovaries that are located in the pelvis. As a result, these patients have well-developed secondary gender characteristics, however, do not have menarche.[2,3] In our case, uterine agenesis with complete vaginal development was observed, in pelvic examination vulva was normal and the depth of vagina was 8 cm, which is within limits of normal female external genitalia with the complete absence of uterus, and both tubes and the ovaries were normal. Also in our case, laboratory findings (FSH, LH, estradiol, prolactin, DHEA-S) were normal.

In the etiology of MRKH syndrome, the nonunion of Mullerian duct with Wolfian duct, and the deficiency in the estrogen and progesterone receptors are blamed. The deficiency blocks the differentiation of embryonic Mullerian duct. Mayer-Rokitansky-Kuster-Hauser syndrome is usually seen together with urinary tract and skeletal system abnormalities. In our case, we have detected right ectopic pelvic kidney, and a perirenal cyst with unknown origin adjacent to pelvic kidney. Oppelt et al[4] in their study defined associated malformation in more than 1/3 of MRKH syndrome cases. Together with this syndrome, renal agenesis, ectopic kidney or absence of a kidney, the duplication of the collecting ducts and the urinary system was seen by 40%. The skeletal system anomalies and they are usually the anomalies of vertebra such as fused cervical vertebrae (Klippel-Feil syndrome), costae and upper extremities can also come along this syndrome. Additionally, skeletal anomalies such as sacralisation, spina bifida, and also congenital aortic aneurysms can be observed.[5] In our case, there was urinary tract anomaly, However, there was not anomalies related to skeletal system. Mayer-Rokitansky-Kuster-Hauser syndrome can be inherited autosomal dominantly, however, frequently is sporadic. The karyotype of patients is usually 46,XX. There were no any problems follow up to our case. We informed the patient that if she has married she would be able to have a normal sexual life, although she would not be able to have a child in a normal way.

In conclusion, the coexistence of genitourinary tract abnormalities should always be kept in mind during the evaluation of primary amenorrheic cases.

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