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CASE REPORT

Vaginal Agenesis Associated with Cervical Hypoplasia and Bicornuate Uterus

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ABSTRACT

A 16-year-old woman presented with primary amenorrhea and progressively cyclic lower abdominal pain. Physical examination revealed well-developed secondary sexual characteristics. Pelvic examination revealed normal appearing external genitalia and an absent vagina. Pelvic sonogram revealed hypoplastic cervix and right adnexal mass. Laparoscopy visualized additional information of bicornuate uterus and severe endometriosis. The operative findings disclosed vaginal agenesis, hypoplastic cervix, bicornuate uterus, right hematosalpinx, right ovarian chocolate cyst, multiple endometriotic spots at cul-de-sac, and severe pelvic adhesion. Hysterectomy and right salpingo-oophorectomy were performed. Left ovary was preserved to maintain hormonal production. Pathological study had confirmed the diagnosis of endometriosis and cervical hypoplasia. Vaginal reconstruction was discussed with the patient. Chromosome study was performed with the result of 46,XX. Clinical features and management of this rare Müllerian anomaly were also discussed.

Key words: vaginal agenesis, cervical hypoplasia, endometriosis

Vaginal agenesis, one of Momalies, is a rare congenital malformation.[1] It is typically accompanied by absent uterus that is known as Mayer-Rokitansky-Küster-Hauser syndrome.[1,2] It is rarer if the uterus is present in a patient with vaginal agenesis. If the uterus is present, it is usually malformed as a rudiment and associated with a non-functioning endometrium.[3,4] Therefore, it is extremely rare if vaginal agenesis is associated with a near completely developed uterus with functioning endometrium.[3,4] The presence of a functioning endometrium combined with vaginal agenesis causes a retrograde menstruation and then a greater likelihood of endometriosis.[4,5] This patient had vaginal agenesis associated with near completely developed uterus and functional endometrium. The patient also suffered from pelvic pain according to this explainable process.

Case report

A 16-year-old woman presented with primary amenorrhea and progressively cyclic lower abdominal pain. She had suffered from cyclic pain for 1 year and her symptom had been progressively severe for 6 months. She had ventricular septal defect of heart with a functional class 1 status. On physical examination, she had normal appearing general appearance, a pansystolic murmur grade 5/6 at left upper parasternal border, no scoliosis of spine, and well-developed

secondary sexual characteristics. On pelvic examination, it revealed normal appearing external genitalia and an absent vagina. On rectal examination, uterus was palpated with suspected right adnexal mass. Pelvic sonogram was then performed, it revealed hypoplastic cervix and right adnexal mass (Fig. 1.). Laparoscopy was performed, it helped visualize additional information of bicornuate uterus and severe endometriosis. Intravenous pyelogram and hearing assessment were performed to rule out associated anomalies, both had normal results. Chromosome study was performed, the result was 46,XX.

After extensive counseling, the patient decided to undergo hysterectomy. The operative findings disclosed vaginal agenesis, hypoplastic cervix, bicornuate uterus, right hematosalpinx (15x4x5 cm), right ovarian chocolate cyst (3x3x3 cm), multiple endometriotic spots at cul-de-sac, and severe pelvic adhesion. Total abdominal hysterectomy and right salpingo-oophorectomy were performed (Fig. 2.). Left ovary was preserved to maintain hormonal production. Pathological study had confirmed the diagnosis of endometriosis and cervical hypoplasia. Vaginal reconstruction was discussed with the patient but she decided not to have that operation.

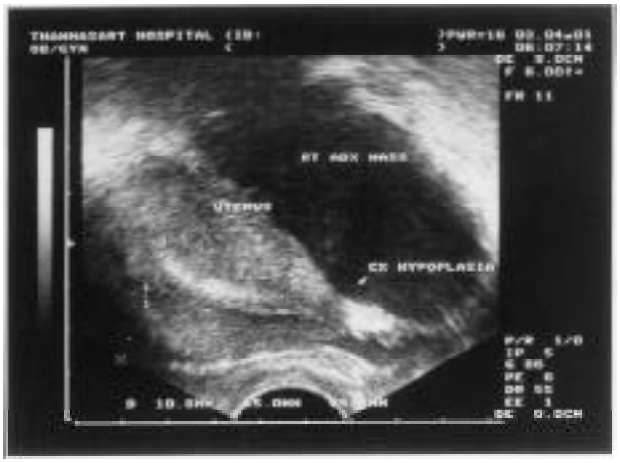

Fig. 1. Ultrasound picture showed a uterus, right adnexal cystic mass (sausage-like), and hypoplastic cervix.

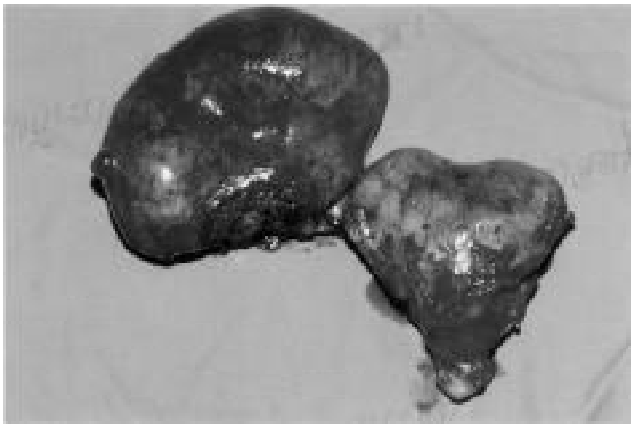

Fig. 2. Gross pathology showed a bicornuate uterus, right hematosalpinx, and hypoplastic cervix.

Discussion

Most patients who present with primary amenorrhea and cyclic pelvic pain in suspicion of outflow tract obstruction are more likely caused by imperforate hymen or transverse vaginal septum.[3] It is important to distinguish these conditions from vaginal agenesis associated with functioning endometrium because the treatment is different for each of them. Pitfall in diagnosis may occur from the matter of fact that vaginal agenesis is mostly associated with absent uterus.[1] If the uterus is present, therefore, the condition should be more likely given a diagnosis of imperforate hymen or transverse vaginal septum. Transverse vaginal septum is particularly possible to be misdiagnosed because imperforate hymen usually is seen externally. Careful ultrasound can aid the diagnosis. Such as in this patient, there was no evidence of hematocolpos that should be found in transverse vaginal septum, but there was evidence of hypoplastic cervix that was more suspicious of Müllerian anomalies (Fig. 1). It is commented that careful physical examination and investigations are essential in diagnosis regardless of the prevalence of the condition. Diagnostic laparoscopy is still a recommended procedure as indicated.[4,6,7] Most recently, 3D ultrasonography and magnetic resonance imaging may be included as confirmatory investigative methods.[8,9] There was a study reported that 3D ultrasound had 91.6 % correlated with the external uterine configuration observed by laparoscopy.[8]

Müllerian anomalies have been found to be associated with other anomalies. The spinal abnormalities are the most common anomalies and include scoliosis, fused, or wedged vertebrae.[10] Renal abnormalities are present in approximately one-third of the cases of Müllerian agenesis and may include renal agenesis, malrotation, or ectopic kidneys.[1] To investigate for associated anomalies is essential as such investigations this patient had.

There are several potential hypotheses of the disorder especially the activating mutation of antimüllerian hormone gene that causes the regression of the Müllerian structures.[11] The etiology for that is unknown. Chromosome abnormalities are not responsible for the process.[12,13] Affected individuals are best explained on the basis of polygenic/multifactorial inheritance.[12,13] There was a recent study showed certain associations between Mayer-Rokitansky-Küster-Hauser syndrome and the major histocompatibility complex.[14] Interestingly, this patient also has a congenital anomaly of the heart that is a ventricular septal defect. From intensive review, there have been no associations between Müllerian anomaly and heart defect.[1,2,6,12,13] There may be 2 possible explanations concerning about the heart defect in this patient. First, it may be sporadic that incidentally occurs in the patient. Second, it may be questioned in term of any association with Müllerian anomaly that may be mentioned for further study. This patient also suffered from pelvic endometriosis. The process of endometriosis in this condition has been thought to be the result of mechanical obstruction.[3,4,5,15]

Management of the patient consists of correction of obstruction and endometriosis, creation of neovagina, and psychological management. In term of menstrual obstruction and endometriosis, hysterectomy remains a proper treatment method.[6,16,17] Ovaries should be remained to maintain hormonal production. Laparoscopic hysterectomy may be an alternative in the present time.[18,19] Creation of neovagina either surgical or non-surgical method must be extensively discussed with the patient. It usually is delayed until the patient is ready to engage in sexual activity so that the patient will be motivated to follow management recommendations.[20] Psychological aspect is crucial as an important part of counseling with the patient and also with the family, the patient is commonly affected on her self-esteem.[21]

In summary, Mayer-Rokitansky-Küster-Hauser syndrome comprises combined hypoplasia of the vagina and the uterus.[1,2] Rare occasion, vaginal agenesis is associated with a near completely developed uterus with functioning endometrium.[3,4] This article has reported this rare case in order to describe some interesting points of the anomalies especially the association between vaginal agenesis and the near

completely developed uterus. Again, careful examination and investigations are essential to avoid the misdiagnosis. Documentation of any associated renal and skeletal anomalies is recommended. Counseling with respect to patient's decision is important including proper psychological preparation of the patient for anticipated corrective vaginal surgery.

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