ISSN 2689-8268 | Volume 7

American Journal of Surgery and Clinical Case Reports

Case Report Open Access

Distal Vaginal Aplasia: About a Case and Review of the Literature

El Hachami.FZ[1] Imami Y[1], Elkarouani D[1], Sakim M[1] Gotni A[2], Boufettal H[3], Mahdaoui S[3] and Samouh N[3]

[1]Gynecology obstetrics residents at CHU IBN ROCHD Casablanca, Morocco

[2]Assistant professor of Gynecology obstetrics at CHU IBN ROCHD Casablanca, Morocco

[3]Professors of Gynecology obstetrics at: Gynecology Department C, CHU Ibn Rochd Casablanca, Morocco

***Corresponding author:**

EL HACHAMI.FZÂ,
Gynecology obstetrics residents at CHU IBN ROCHD Casablanca, Morocco

Received: 02 Jan 2024
Accepted: 27 Jan 2024
Published: 02 Feb 2024
J Short Name: AJSCCR

Keywords:

Hematocolpos; Imperforate hymen; Distal vaginal agenesis; Vaginoplasty; Vaginal stretchin

Citation:

El Hachami.FZÂ. Distal Vaginal Aplasia: About a Case and Review of the Literature. Ame J Surg Clin Case Rep. 2024; 7(9): 1-4

1. Abstract

Congenital anomalies of the genital tract are rare and present with a broad spectrum of clinical features, making their diagnosis a challenging process. Vaginal aplasia is a rare congenital malformation whose classic clinical picture is that of primary amenorrhea. Diagnosis is based on clinical examination and imaging. The treatment of distal vaginal agenesis is exclusively surgical. Comprehensive management is imperative to preserve fertility and functionality.

2. Introduction

Isolated distal vaginal atresia is a rare Müllerian anomaly characterized by development of the uterus, cervix and upper vagina, with an atrophied lower vaginal segment. This condition frequently manifests itself at puberty, when an obstructed menstrual cycle causes hematometrocolposis. Patients often present with acute abdominal pain and, on evaluation, may present with an abdominal mass and lack of vaginal opening. The most effective imaging remains pelvic magnetic resonance imaging [MRI], which can confirm the absence of the distal part of the vagina as well as assess the urinary tract, given the frequent association of urinary tract anomalies with mullerian anomalies, and aid preoperative planning by estimating the distance between the vaginal bulge and the perineum.

The aim of this work is to analyze the epidemiological characteristics, clinical, evolutionary and therapeutic outcomes, through a case study of distal vaginal aplasia at the Mohammed VI center service of the University Hospital of Casablanca.

3. Identity

The patient was 21 years old, with no particular pathological antecedents, and presented with a primary amenorrhea with no accompanying signs. Clinical examination revealed an impermeable hymen with rectal examination and abdominal palpation revealed an enlarged uterus reaching midway to the umbilicus, with a perceived posterior bulge. Abdominal and pelvic ultrasonography revealed an enlarged anteverted uterus with a discreetly half-open endocavitary line upstream of an endovaginal collection occupying the entire vaginal cavity, with a liquid-liquid level measuring 7.5x3 cm, the site of hyperechoic declivity [hemosiderin deposition]. The ovaries, kidneys and bladder were normal. MRI revealed an enlarged anteverted uterus of normal architecture, with vaginal distension in T1 hyper signal, T2 hypo signal and distal vaginal atresia. Within this vaginal distension, small declining formations in signal void on all sequences were associated with hemosiderin deposits. This collection continues superiorly with moderate upstream endocavitary retention in T1 hyper signal.

Signal integrity and cervical thickness were present, the rest of the MRI was unremarkable. The patient underwent an examination under general anaesthetic for a possible hematocolpos cure, which revealed an inability to explore the vagina due to the complete absence of the orifice in its lower part. The patient will be scheduled for reconstruction surgery (Figure 1-3).

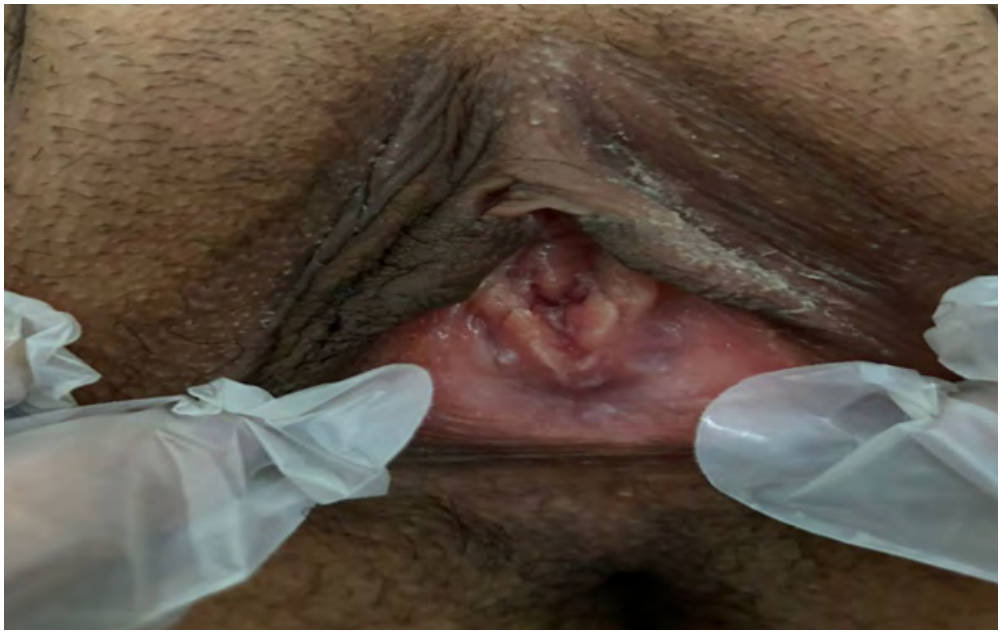

Figure 1: Appearance of the introitus in a patient with distal vaginal agenesis

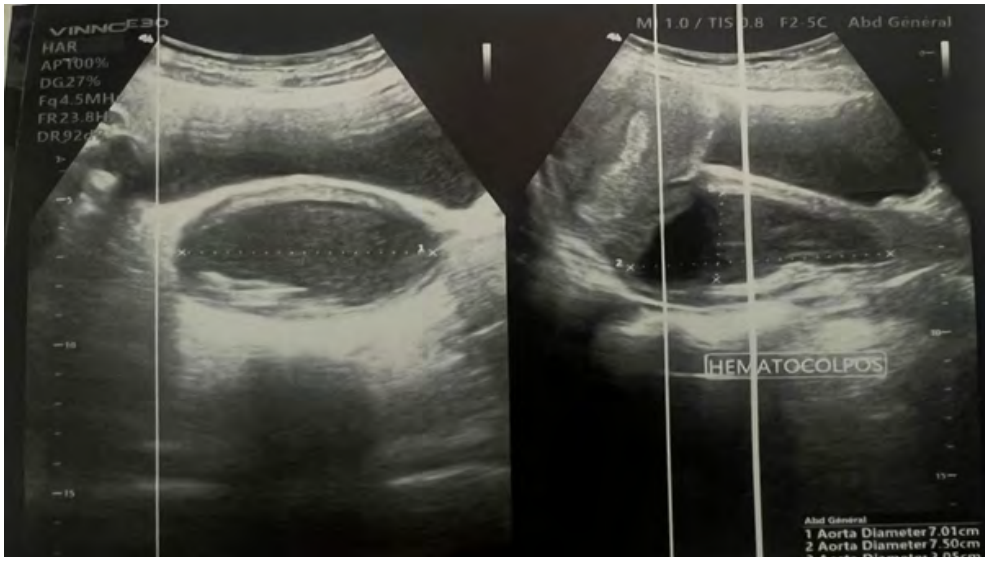

Figure 2: Ultrasonographic appearance of a 7.5×3 cm hematocolpos

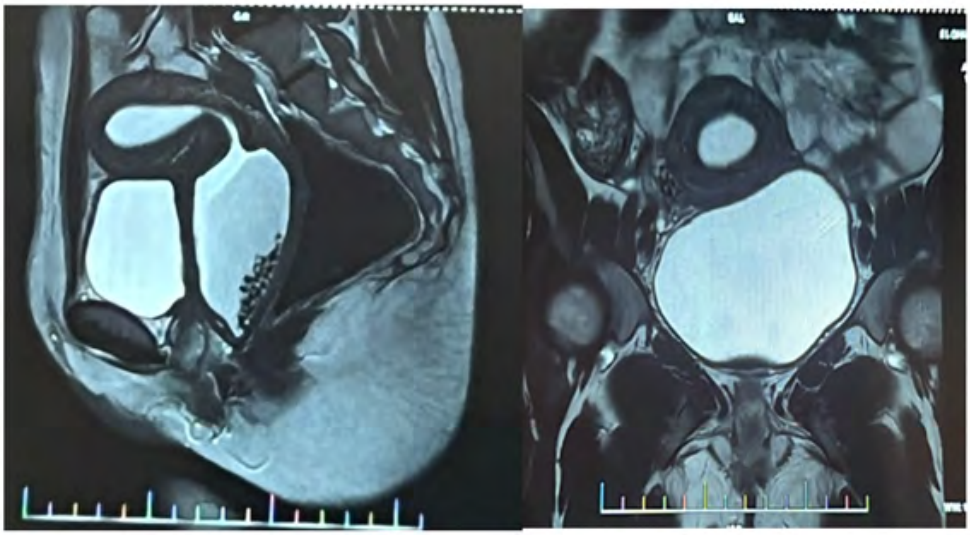

Figure 3: Appearance of an 11x5.5 cm hematocolpos with moderate upstream hematometry. [(A) ; (B) - section]

4. Discussion

Hematocolpos is a medical condition in which menstrual blood or secretory fluid accumulates in the vagina due to vaginal obstruction. Hematocolpos can occur as a result of congenital urogenital anomalies or acquired vaginal occlusion. Hematocolpos due to vaginal agenesis with functional uterus is particularly rare [12, 13]. There are four main congenital causes of hematocolpos: imperforate hymen, distal vaginal agenesis, complete transverse vaginal septum, obstructed hemivagina and homolateral renal anomaly [OHVIRA]. Vaginal agenesis is classified as complete, proximal and distal agenesis. Congenital agenesis of the lower vagina is a rare condition estimated at 5% of the total number. The prevalence of vaginal agenesis ranges from one in 4,000 to one in 10,000 women [6]. Various theories have been put forward. The most widely accepted theory is that the upper 2/3 of the vagina is developed from the Müllerian while the urogenital sinus contributes to the lower 1/3 of the vagina [2].

As the lower vaginal segment develops from the urogenital sinus, distal vaginal agenesis is the inability of the urogenital sinus to form the lower vaginal segment, or the partial inability of the vaginal plate to canalize. According to the American Society for Reproductive Medicine [ASRM] classification, distal vaginal agenesis is classified as a class IA anomaly [9, 16]. Our knowledge of the genes involved in vaginal development comes from studies of genetic syndromes with vaginal abnormalities, or from knock-out mouse models. Both types of study have identified key genes regulating vaginal development. To date, only one study has reported TBX6 as a candidate causing distal vaginal atresia by exome sequencing. Knockout mouse models have verified several molecular factors essential for Müller duct formation and development. Spontaneous point mutation of mouse Lhfpl2 leads to an abnormal upper longitudinal vaginal septum and distal vaginal agenesis, with a normal ovary and uterus . Female mice lacking the Tyro3 RTK subfamily [Tyro3 , Axl and Mer] have a high incidence of distal vaginal atresia [2]. The application of genomic sequencing [GS] in clinical research has led to the discovery of many genomic variations in genetic diseases.

Diagnosis can be made with hydrocolpos on antenatal or post-natal ultrasound, though more common diagnosis is made during adolescence with symptoms of primary amenorrhea, cyclic abdominal pain, and sometimes an abdominal mass. Endometriosis and pelvic adhesions also can occur due to retrograde menstruation [4, 5]. Clinically, distal vaginal atresia presents similarly to an imperforate hymen and a transverse vaginal septum. Perineal inspection reveals absence of hymen and vaginal orifice; however, with a small concave dimple [6, 7].The diferentiation is imperative for proper surgical planning [8]. Perineal inspection reveals the absence of hymen and vaginal orifice; however, with a small concave dimple. Depressibility and depth of the vaginal cup can vary from almost nothing to a few centimetres. One of the advantages of the initial examination is to assess the possibility of creating a neo-vagina. In patients with hematocolpos, transabdominal ultrasound may reveal the distended upper vagina with moving internal echoes, and ultrasound with transperineal approach may demonstrate the length of the atresic vaginal segment. The uterus and cervix may be normal or show defects in fusion or resorption, while the distal or total vagina is replaced by fibrous tissue [2]. Although distal vaginal agenesis can also be suggested by CT scan, MRI allows better delineation of the atresic vaginal segment, enabling differential diagnosis of the more frequent hematocolpos due to imperforate hymen. MRI is essential for morphological evaluation and surgical planning of distal vaginal agenesis [graft length adjustment] [1]. Vaginal atresia is classified as U0-4/C0-3/V4 according to the new European Society of Human Reproduction and Embryology/European Society of Gynecological Endoscopy [ESHRE/ESGE] classification system for female genital anomalies [3].

The treatment of distal vaginal agenesis is exclusively surgical. A local traction vaginoplasty with direct anastomosis of the superior vaginal mucosa to the introitus via a perineal incision is performed when the distance between the perineal surface and the caudal aspect of the distended vagina is 2 cm or less [1]. Due to the absence of a large vaginal segment, local traction vaginoplasty is difficult to perform as it results in shortening of the vagina, especially when the thickness of the agenesic segment is ≥2 cm [10, 15]. Vaginoplasty using skin or bowel grafts has been performed when a large segment of the vagina is absent [1]. The timing of surgery is controversial. Cure should be deferred until the appearance of hematocolpos so that dilated upper vaginal segments are larger, facilitating determination of the best surgical incision route and reduction of graft length [1, 11]. In cases of greater agenesis and risk of vaginal stenosis postoperatively, replacement of the missing part of the vagina with other tissues or a modified balloon vaginoplasty may be used. Recurrent vaginal stenosis is the most frequent late complication, and follow-up is essential to prevent mild vaginal stenosis after the operation and to confirm the absence of tension on the vaginal mucosa or suture line. An inflatable vaginal stent with an internal drain coated with an estrogen-based ointment, inserted at night until the end of the constrictive phase of healing, has been used to prevent stenosis [10]. The treatment of vaginal aplasia consists in creating a neo-vagina, with the aim of facilitating the onset of sexual life.

5. Conclusion

Vaginal aplasia is a rare congenital malformation whose classic clinical picture is that of primary amenorrhea with chronic cyclical pelvic pain. Diagnosis is based on clinical examination and imaging. MRI is used to assess the extent of atresia, and to guide surgical management. Whatever surgical technique is used, its aim is to restore the integrity of the utero-vaginal tract, thus enabling these patients, whose psychological experience is particularly difficult, to enjoy a satisfying sex life and the possibility of pregnancy.

References

1. Tanitame K, Tanitame N, Urayama S, Ohtsu K. Congenital anomalies causing hemato/hydrocolpos: imaging fndings, treatments, and outcomes. 2021.
2. Kang J, Zhou, Chen N, Liu Z, Zhang Y, Sun J, et al. Clinical and Genetic Characteristics of a Cohort with Distal Vaginal Atresia. Int J Mol Sci.2022; 23(21): 12853.
3. Grimbizis GF, Gordts S, Di Spiezio Sardo A, Brucker S, De Angelis C, Gergolet M, et al. Le consensus ESHRE/ESGE sur la classification des anomalies congénitales de l'appareil génital féminin. Hum. Reproduction. 2013; 28: 2032-2044.
4. Damaris F, Liselotte M, Nicolai M, Veronika G, Ibrahim A. Anomalies utérines et endometrioses.
5. Sanfilippo JS, Wakim NG, Schikler KN, Yussman MA. Endométriose en association avec une anomalie uterine. Suis. J.Obstet. Gynécol. 1986; 154: 39-43.
6. Tanitame K, Tanitame N, Urayama S, Ohtsu K. Congenital anomalies causing hemato/hydrocolpos: imaging findings, treatments, and outcomes.
7. Ugur MG, Balat O, Ozturk E, Bekerecioglu M, Dikensoy E. Pitfalls in the diagnosis and management of distal vaginal agenesis: 10 years of experience in a single center. Eur J Obstet Gynecol Reprod Biol. 2012; 163: 85-90.
8. Meklaa A, Moussaoui DR, Dehayni M. Gynecology-obstetrics department, the Mohamed V Rabat military training hospital.
9. The American Fertility Society. The American Fertility Society classifcations of adnexal adhesions, distal tubal occlusion, tubal occlusion secondary to tubal ligation, tubal pregnancies, müllerian anomalies and intrauterine adhesions. Fertil Steril. 1988; 49: 944–55.
10. Ugur MG, Balat O, Ozturk E, Bekerecioglu M, Dikensoy E. Pitfalls in diagnosis and management of distal vaginal agenesis: 10-year experience at a single centre. Eur J Obstet Gynecol Reprod Biol. 2012; 163: 85–90.
11. Dural O, Ugurlucan FG, Yasa C, Bastu E, Eren H, Yuksel B, et al. A case of distal vaginal agenesis presenting with recurrent urinary tract infection and pyuria in a prepubertal girl. J Pediatr Adolesc Gynecol. 2017; 30: e7–e10.
12. Grimbizis GF, Camus M, Tarlatzis BC, Bontis JN, Devroey P. Clinical implications of uterine malformations and results of hysteroscopic treatment. Hmm. Reproduction. Update 2001; 7:161–74.
13. Ugur MG, Balat O, Ozturk E, Bekerecioglu M, Dikensoy E. Pitfalls in the diagnosis and management of distal vaginal agenesis: 10 years of experience in a single center. EUR. J. Obstet. Gynecol. Reproduction. Biol. 2012; 163:85–90.
14. Imai K, Aoyama K, Goto T, Kitaori T, Iguchi T, Sasano H, et al. Hematocolpos due to lower vaginal agenesis in an adolescent girl Acute medical surgery. 2023; 10(1): e832.
15. Tercan M, Balat O, Bekerecioglu M, Atik B. The use of fibrin glue in the McIndoe technique of vaginoplasty. Plastic and Reconstructive Surgery. 2002; 109: 706-709.
16. Ismail HH, Yonis R, Asiri A, Aslan M, Ibrahim W. Distal Vaginal Atresia: Bahrain Med Bull. 2020; 42 (4): 313 – 315.