The Review Of
DIABETIC
STUDIES OPEN ACCESS

Unicornuate Uterus With Non-Communicating Functional Rudimentary Horn In Association With VACTERL Spectrum: Case Report

José Luis Granizo Jara[1], Juan Carlos Sánchez Solis[2], Mayra Raquel Ortiz Andaluz[3], Gabriela Alexandra Granizo Jara[4], Pamela Nayely Sánchez Silva[5], Lisbeth Josefina Reales Chacon[6], Jonathan Alexander Lizano Caguana[7], Danilo Javier Orozco Gaguancela[8]

[1]*Universidad Nacional de Chimborazo, Riobamba – Ecuador*
Email: *jluisgj24@hotmail.com*
ORCID: *https://orcid.org/0009-0003-7855-0838*
[2]*Ambato – Ecuador* Email: *drjuanks@hotmail.com*
ORCID: *https://orcid.org/0009-0008-3206-6422*
[3]*Universidad Tecnológica Indoamérica, Ambato – Ecuador*
Email: *mayraortiz@uti.edu.ec*
ORCID: *https://orcid.org/0009-0008-1932-9371*
[4]*Riobamba – Ecuador*
Email: *alexagj22@hotmail.com*
ORCID: *https://orcid.org/0009-0002-9641-9529*
[5]*Universidad Nacional de Chimborazo, Riobamba – Ecuador*
Email: *nayely.sanchez@unach.edu.ec*
ORCID: *https://orcid.org/0009-0006-6860-3635*
[6]*Universidad Nacional de Chimborazo, Riobamba – Ecuador*
Email: *lisbeth.reales@unach.edu.ec*
ORCID: *https://orcid.org/0000-0002-4242-3429*
[7]Email: *jonathanlizano2013@gmail.com*
ORCID: *https://orcid.org/0000-0003-1619-0518*
[8]*Universidad Nacional de Chimborazo*
Email: *danymilos@hotmail.com*
ORCID: *https://orcid.org/0009-0003-4188-5335*

Abstract

The unicornuate uterus with a non-communicating rudimentary horn (UUNCRH) is one of the least common congenital uterine malformations. It is defined as a "hemiuterus" with adnexa, whose affected contralateral side develops as a vestige of hollow fibromuscular tissue that does not communicate with the main uterine or vaginal cavity, but may retain functional endometrium. Recently, a close relationship has been demonstrated between UUNCRH and other congenital malformations described in the VACTERL spectrum, a set of vertebral, anorectal, cardiovascular, tracheoesophageal, renal, and limb defects. This link has been documented only twice in the world, therefore, we present the first case in America and Ecuador of a 34-year-old patient with a history of congenital vertebral, anorectal, and renal malformations, diagnosed with UUNCRH. This case highlights several inconsistencies regarding the diagnostic protocol applied, because, despite the fact that the patient had previously undergone several imaging studies and gynecological-obstetric surgical procedures, the congenital defect was not identified in time, which emphasizes the importance of including genital anatomical screening in patients with a history of other congenital structural anomalies.

Key words: Unicornuate uterus, congenital uterine malformations, hemi-uterus, VACTERL spectrum, Genital anatomical screening, gyneco-obstetric interventions.

Introduction

The unicornuate uterus is one of the least frequent uterine malformations, it occurs in 0.1-0.5% of the general population, of which 74-90% are associated with the rudimentary horn variant and 25% of them are non-communicating, thus, the unicornuate uterus with a non-communicating functional rudimentary horn (UUNCRH) is defined as a "hemiuterus" with annexes, whose affected contralateral develops as a vestige of hollow fibromuscular tissue, which does not maintain communication with the main uterine or vaginal cavity, however, it can retain functionality by lining the endometrium. (1)(2)

However, its etiology is attributed to a defect in the fusion of the Müller's or paramesonephric ducts during intrauterine development and is usually asymptomatic until before menarche or even after, in fact, 78% of cases are detected in the third decade of life as a fortuitous finding in the practice of imaging studies, or by complications, which manifest themselves with intense abdominal or pelvic pain, amenorrhea, dysmenorrhea, hematocolpos, dyspareunia, infertility, endometriosis, miscarriage, ectopic pregnancy, preterm delivery, intrauterine growth retardation, among others. (3) (3)(4)(5)

Recently, a close relationship has been evidenced between UUNCRH and other congenital malformations, several of them described in the VACTERL association, which includes a set of vertebral, anorectal, cardiovascular, tracheoesophageal, renal and extremity defects that can manifest simultaneously at birth and is characterized by the presence of at least three of them. Although it is hypothesized that congenital anomalies of the VACTERL spectrum may be predictors of gynecological disorders, there is still a limited field of study about this relationship.(6)(7)

To date, there are only 2 documented cases in the world, which link UUNCRH with VACTERL association, which emphasizes the importance of enriching the bibliographic base available through the report of new cases that allow identifying predictive patterns or variables of this pathology and supporting or refuting the proposed theory, which represents a great contribution to the early diagnosis of uterine malformations. and the standardization of timely therapeutic behaviors.(8) (9)

In this context, we present the case of a 34-year-old patient with a history of congenital vertebral, anorectal and renal malformations, diagnosed with UUNCRH after exploratory laparoscopy for a right pelvic mass of unknown cause. The objective of this study is to document the third case in the world and the first in America and Ecuador, of a unicornuate uterus with a rudimentary non-communicating functional horn, associated with the VACTERL spectrum.

Case Report

A 34-year-old female, born and resident in Ambato-Ecuador, with a congenital history of imperforate anus (AI), scoliosis, hip dysplasia and right renal agenesis (RA), during her adolescence is diagnosed with antiphospholipid syndrome and rheumatoid arthritis, underwent correction of the imperforate anus at birth, dorsal spinal arthrodesis, appendectomy and right hip arthroplasty.

Menarche at 13 years of age, irregular and painful menstruation, subdermal implantation and subsequent salpingectomy as contraception, a procedure in which didelphic uterus is diagnosed. 1 abortion, 0 births, 3 cesarean sections. As a result of the first pregnancy, female newborn, born at 32 weeks, was admitted to neonatology, died on the third day; second pregnancy, male newborn, 29 weeks, admission to neonatology, alive; third male newborn pregnancy, 27 weeks, admission to neonatology, alive.

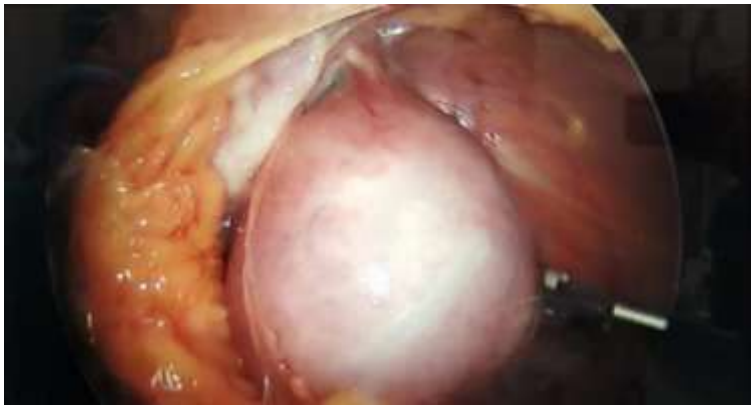
The patient presented with menorrhagia and intense abdominal pain, the latter located in the iliac fossa and right flank, with a real onset date in 2023 being occasional and tolerable, however, a year later the pain intensified to 10/10 on the VAS scale, exacerbating during menstruation, accompanied by sporadic emesis, the patient self-medicated NSAIDs without remission of the

condition. On physical examination, patient in poor general condition, algic facie, painful abdomen on superficial and deep palpation in the iliac fossa and right flank, apparent mass in the right pelvic area was palpated, active vaginal bleeding.

Complementary laboratory and imaging tests are requested. The transvaginal ultrasound report concludes that a right ovarian cyst with possibilities of malignancy is discarded, which is ruled out with negative tumor markers, subsequently, an abdominal tomography is requested that reports left iliac adenopathies of etiology to be determined and an increase in the right ovarian volume, which after clinical analysis is adjudicated diagnostic suspicion of complex cyst, however, as it does not have conclusive results and due to lack of remission of the picture, Despite the establishment of an analgesic scheme, it was decided to perform exploratory laparoscopy.

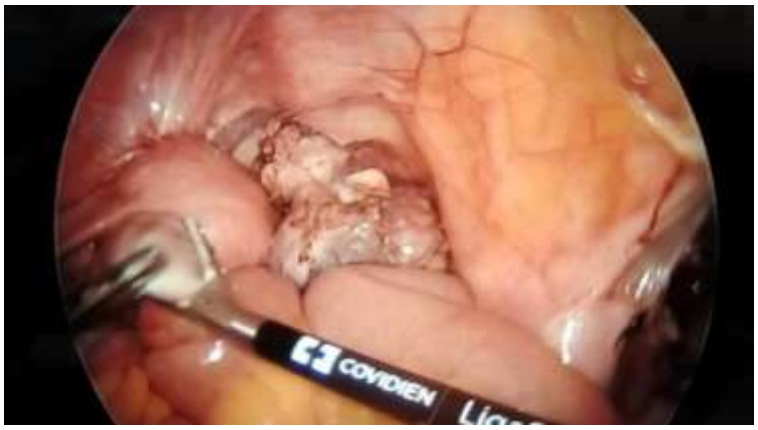
With the necessary measures adjusted to the patient's comorbidity condition, surgical intervention was performed, showing multiple adhesions in the right iliac fossa, then, during the pelvic examination, a simple cyst of the left ovary was observed, however it was not possible to find a fallopian tube or right ovary, when adhesiolysis was performed, a large mass was found in the right iliac fossa, compatible with uterine and ovarian adnexa with functional endometrium, which produces hematometry of 200cc, drainage and subsequent rudimentary horn hysterectomy are performed. Patient in immediate and immediate postoperative period with adequate clinical evolution.

Figure 1 Right uterine remnant with right fallopian tube and ovary, covered by omentum.



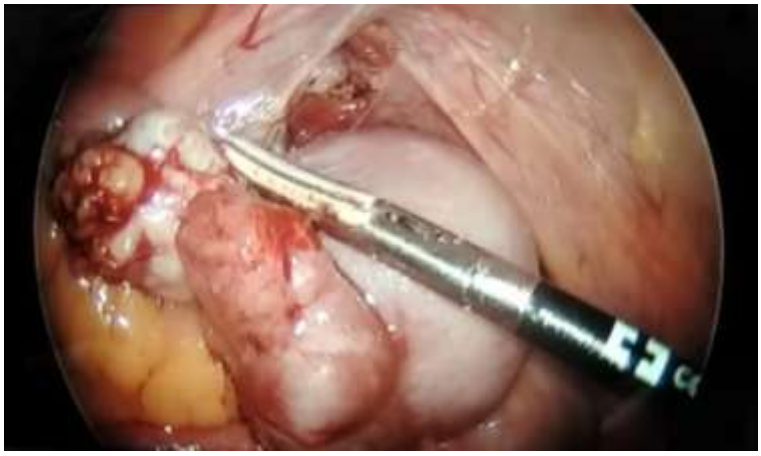
Own source.

Figure 2 Left unicornuate uterus with left fallopian tube and ovary, at the level of the fundus of the Douglas sac.



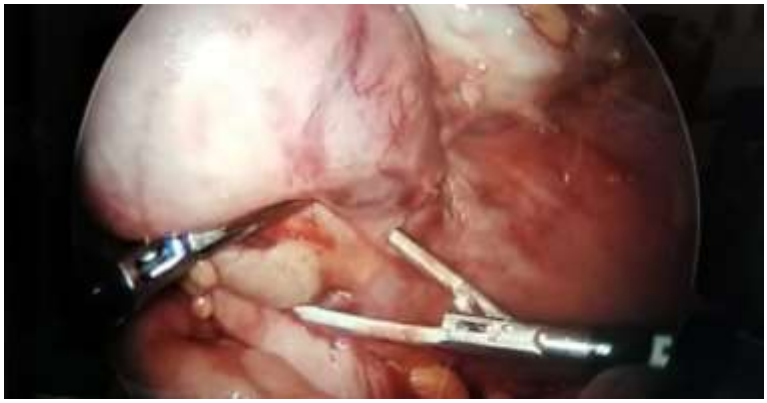
Own source.

Figure 3 Section of the tube and ovary of uterine remnant.



Own source.

Figure 4 Section of rudimentary uterus.



Own source.

Discussion

The relationship between UUNCRH and VACTERL association has been described only twice in the world, the first case was registered in Europe in 2009, corresponding to a 28-year-old nulliparous woman with a history of vertebral aplasia, AI, anovaginal fistula, right RA and polydactyly, diagnosed with a unicornuate uterus with a rudimentary horn, later, in 2019, a second case is reported in Asia, it is a 17-year-old girl, nulliparous, with the same diagnosis and medical history of AI, tracheoesophageal malformation and left RA. Under this perspective, the ongoing research presents the third case in the world and the first in the American continent that relates the two spectrums.(8)(9)

The link between these congenital anomalies could be embryologically justified, since both the female genital system and the urinary system share the same origin in the intermediate mesoderm and in principle, the excretory systems of the two tracts lead to a common structure called cloaca. During the sixth week of gestation, the female genital tract is formed with the growth of the Müller's ducts, these are medially fused into a single structure joined by a septum, which must be reabsorbed between 12 and 20 weeks, giving rise to fallopian tubes, uterus, cervix and upper 2/3 of the vagina. Therefore, when one of these ducts develops correctly, while its contralateral evolves partially, the result is a unicornuate uterus with a rudimentary horn. (10)(5)(2)

On the other hand, the development of the female urinary tract begins around the fourth week, through 3 sequential renal systems, from cranial to caudal: pronephros, mesonephros and metanephros, the first two are transient. The mesonephros drains the urine into the cloaca through Wolff's collecting ducts that subsequently involute in the woman, meanwhile, the definitive

kidney (metanephros) emerges from the metanephrogenic blastema and the ureteral bud, a branch of this same duct. Between the fourth and seventh week the cloaca divides to form the anterior urogenital sinus (lower vagina, bladder, urethra) and the posterior anal canal. Thus, this proximity and the simultaneous development of the two tracts could respond to the coexistence of the malformations under study.(10)

Based on the above, several authors have highlighted the importance of this correlation, including Hira Ahmad et al., who, through a retrospective cross-sectional analysis in pediatric patients, point out that findings compatible with VACTERL syndrome (especially anorectal and renal malformations) are linked to an increased risk of uterine anomalies, which should lead to a complete evaluation of the female genital tract. This criterion is shared by Hambraeus and others, who propose the inclusion of the letter "G" (for genital) to the acronym VACTERL, with the aim of early diagnosis of reproductive anomalies.(11)(12)

In accordance with the above, Frances et al., through a retrospective cohort study in women aged 12 to 35 years, show a strong relationship between Mullerian defects and renal anomalies, particularly renal agenesis, agreeing that after the diagnosis of congenital RA, a routine pelvic ultrasound study should be performed around the age of expected menarche. however, new guidelines for the management of patients with RA suggest its application during the first two weeks of life, attributed to the uterine enlargement of the newborn due to the estrogenic hormonal action of the mother.(13)(14)

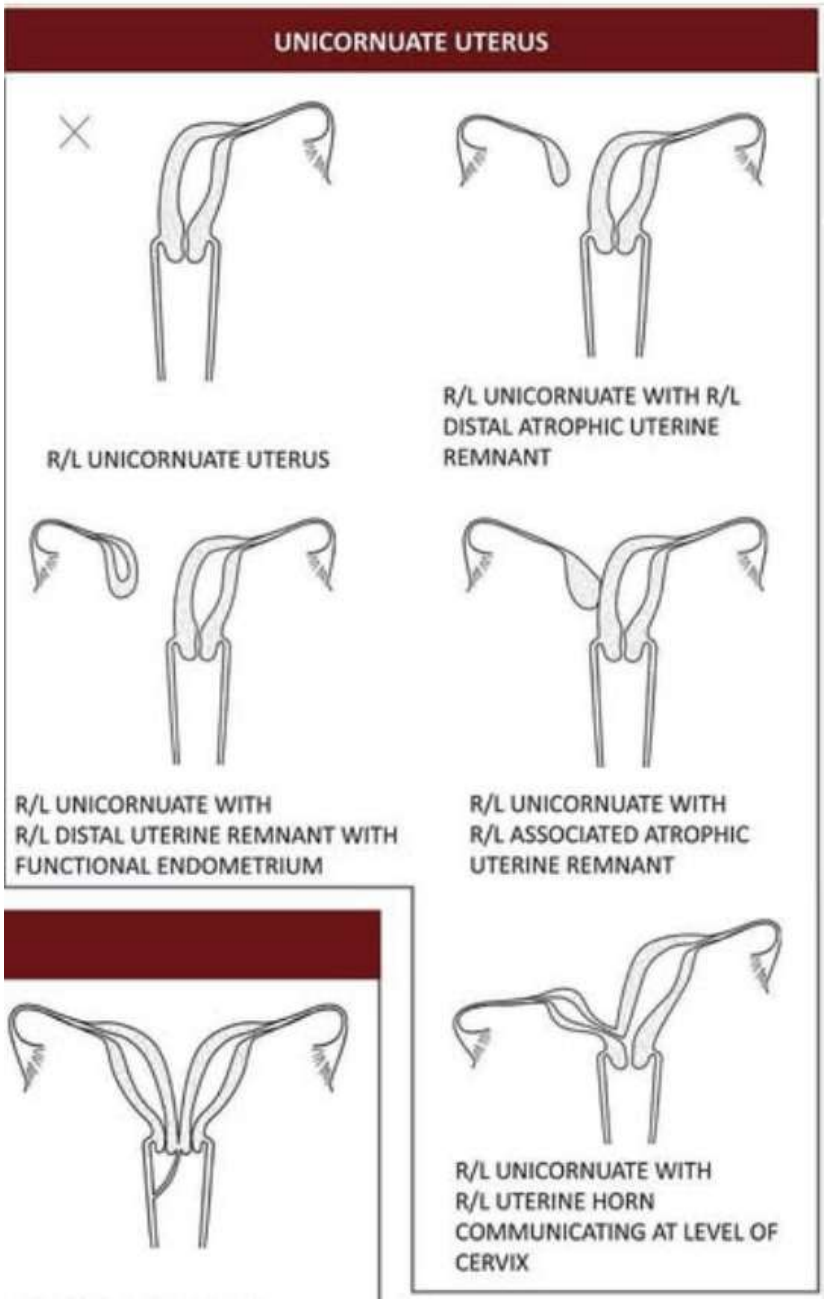
In relation to the case described, several questions arise regarding the diagnostic protocol applied, because despite the fact that the patient was previously subjected to several imaging studies and gynecological-obstetric surgical interventions, the congenital defect was not identified in time. While it is true that conventional 2D ultrasound is the method of choice in the screening of uterine malformations, Jayaprakasan and Ojha indicate that it is not necessarily the most sensitive, especially in UUNCRH, this attributed to its visualization limited to two-dimensional planes, in fact, Bermejo and others affirm that only magnetic resonance imaging (MRI) and 3D ultrasound (US3D) have the ability to assess the uterine cavity and fundus, simultaneously, which in a certain way, justifies the diagnostic omission committed.(4)(15)

Now, the woman had 2 cesarean sections previously performed, where the malformation was not evidenced either, to date, there is almost no scientific evidence that excuses this terrible non-observance, however, in 2024 Mohammed and others report the case of a 25-year-old patient, multigravid, with 2 previous cesarean sections, who undergoes exploratory laparoscopy that identifies an 18-week pregnancy in a rudimentary uterine horn, also evidencing scarring from previous procedures in the lower segment of the unicornuate uterus, according to the authors, these errors can be attributed to the insufficient experience and training of doctors who perform these procedures in countries with scarce health resources and at the same time, emphasize the importance of thoroughly verifying the uterine anatomy and its appendages during cesarean sections (16)

Returning to the central issue, the patient under study underwent a salpingectomy during the third cesarean section, whose operative protocol concludes "unspecified congenital malformation of the uterus and cervix", despite identifying an anomaly, the two tubes are ligated without investigating in depth, and the relatives are erroneously informed of the finding as a didelphic uterus. revealing a series of inconsistencies in the clinical and surgical management of this patient. Subsequent complications made it possible to clarify the definitive diagnosis as Left unicornuate uterus with right distal uterine remnant with functional endometrium, a name that best suits the case based on the classification of uterine malformations designed by the American Society for Reproductive Medicine (ASRM) in 2021. (17)

Figure 5 Classification of uterine anomalies for unicornuate uterus, according to ASRM 2021.

Fountain:. (17)



Conclusion

The unicornuate uterus with a non-communicating functional rudimentary horn is a very rare congenital uterine malformation, which has been related to the VACTERL spectrum only 2 times in the world, as far as is known, the case presented is the third and the first in America and Ecuador, a precedent that highlights the importance of documenting similar cases that serve as a point of reference and support for the inclusion of genital anatomical screening. in patients with a history of congenital anomalies such as renal agenesis and imperforate anus, allowing timely diagnosis and standardization of the management of these pathologies and reducing complications during the reproductive age. On the other hand, the case under study reveals a series of inconsistencies in the clinical and surgical management of the patient, emphasizing the relevance of expanding the diagnostic possibilities of uterine malformation in women with a history of repeated obstetric complications.

BIBLIOGRAPHY

1. Candiani M, Ciappina N, Fedele F, Bergamini V, Alessandro F. Laparoscopic removal of unseparated cavitated horn in unicornuate uterus: surgical aspects and long-term follow-up. International Journal of Fertility and Sterility. 2023; 17(2): 145-150.

2. Fernández G, Molina R, Montiel N, Fernández M, Oberto J, Velásquez N. Unicornuate uterus. Journal of Obstetrics and Gynecology of Venezuela. 2015; 75(2): 122-131.

3. Fernández M, Pardo M, Leal E, María G, Moral J. Unicornuate uterus with rudimentary horn non-communicator in adolescence. Progress in Obstetrics and Gynecology. 2012; 55(3): 146-149.

4. Jayaprakasan K, Ojha K. Diagnosis of Congenital Uterine Abnormalities: Practical Considerations. Journal of Clinical Medicine. 2022; 11(1251).

5. Chamorro J, Sánchez J, Gómez M. Unicornuate uterus with rudimentary non-communicating cavitated horn: characterization by magnetic resonance imaging. Medical Journal of the Mexican Institute of Social Security. 2018; 56(1): 92-7.

6. Van de Putte R, Van I, Marcelis C, Guo M, Brunner H. Spectrum of congenital anomalies among VACTERL cases: a EUROCAT population-based study. Pediatric Research. 2020; 87(3): 541-549.

7. Hira A, Wood R, Avansino J, Kirsten K. Does presence of a VACTERL anomaly predict an associated gynecologic anomaly in females with anorectal malformations?: A Pediatric Colorectal and Pelvic Learning Consortium Study. Journal of Pediatric Surgery. 2023; 58(3): 471-477.

8. Nunes N, Karandikar S, Cooper S, Jaganathan R, Irani S. VATER/VACTERL syndrome (vertebra/anus/cardiac/trachea/esophogus/radius/renal/limb anomalies) with a noncommunicating functioning uterine horn and a unicornuate uterus: a case report. Fertility and Sterility. 2009; 91(5).

9. Obeidat R, Aleshaw A, Tashtush N, Alsarawi A. Unicornuate uterus with a rudimentary non-communicating cavitary horn in association with VACTERL association: case report. BMC Women´s Health. 2019; 19(71).

10. Azwan R, Utama B, Yusrawati. Urogenital System for Women. Journal of Midwifery. 2021; 6(1).

11. Ahmad H, Wood R, Avansino J, Calkins C. Does presence of a VACTERL anomaly predict an associated gynecologic anomaly in females with anorectal malformations?: A Pediatric Colorectal and Pelvic Learning Consortium Study. J Pediatr Surg. 2023; 58(3): 471-477.

12. Hambraeus M, Börjesson A, Ekmark A, Tofft L, Arnbjörnsson E, P. S. Genital Malformations in Children With VACTERL - Has Time Come to Include "G" in the Acronym? J Pediatr Surg. 2024; 59(10).

13. Frances Y, Smith Y, Wan J, Dendrinos M. Should we screen for Müllerian anomalies following diagnosis of a congenital renal anomaly? Journal of Pediatric Urology. 2022; 18(5): 676.e1-676.e7.

14. Walawender L, Santhanam N, Davies B, Y F. Müllerian anomalies in girls with congenital solitary kidney. Pediatric Nephrology. 2024; 39: 1783–1789.

15. Bermejo C, Puente J, Nicolau B, Alcázar J. Ultrasound diagnosis of uterine malformations and abnormalities of the lower genital tract. Official Journal of the Spanish Society of Gynecology and Obstetrics. 2021; 64: 94-105.

16. Mohammed A, Abdelfattah A, Ali M, Elmubarak L. Intact non-communicating rudimentary horn pregnancy in a patient with a. Women's Health. 2024; 41.

17. Pfeifer S, Attaran M, Goldstein J, Lindheim S. ASRM müllerian classification 2021. Fertility and Sterility. 2021; 116(5): 0015-0282.

18. Grimbizis G, Gordts S, DiSpiezio A, Brucker S. The ESHRE/ESGE consensus on the classification of female genital tract congenital anomalies. Human Reproduction. 2013; 28(8): 2032–2044.

19. Jayaprakasan K, Ojha K. Diagnosis of Congenital Uterine Abnormalities: Practical Considerations. Journal of Clinical Medicine. 2022; 11(5): 1271.

20. Sadek S, Ahmad R, Atia H. Performance of the ESHRE/ESGE classification in differentiating anomalies of double uterine cavity. Middle East Fertility Society Journal. 2016; 21: 75–81.

21. Gaspar J, Ferreira N, Luisa L, Simão C. Unilateral renal agenesis and uterine anomalies:. Birth and Growth Medical Journal. 2017; 26(3).