Open Access

Annals of Surgical Case Reports & Images

Volume 1 | Case Report

Herlyn-Werner-Wunderlich associated hematometra mimicking endometrioid neoformation: A rare case report

***Corresponding Author:** Marco Cerbone*

Email: marcocerbone@gmail.com

Abstract

In this case report we described a a case of ultra-rare Mullerian anomaly associated with renal agenesia in a syndromic framework of Herlyn-Werner-Wunderlich syndrome in a 15 years old patient who complained dysmenorrhea and abdominal pain. Abdominal MRI showed a 18 cm hourglass neoformation with dysomogeneous content, suspected for endometrioma. However, transrectal ultrasound showed the presence of didelphys uterus, a single vagina and two cervices. After a pre-operatory echo-guided operative hysteroscopy, that was unable to drain the hematometra because of the presence of a deep fibrotic septum, a mini-Pfannensteil abdominal laparotomy was performed. The hematometra was drained by hysterotomy and the septum was excised trought the cervix to create a continuity with the vagina. The post-operative period was uneventful, and the patient is now under regular follow-up. We underline the need of multidisciplinary counselling for those complex cases where the genetician, gynecologist, urologist should work together for the need and expectations of the patients.

Keywords: Herlin-weber-wunderlich syndrome; Hematometra; Endometrioid neoformation; Case report.

Luca Maria Schönauer[1,2]; Marco Cerbone[1]*; Maria Bentivoglio[1]; Miriana de Nicolo[1]; Maria Matteo[3]; Antonella Vimercati[1,2]; Ettore Cicinelli[1,2]; Marco Marinaccio[1,2]

[1]Department of Biomedical Sciences and Human Oncology, Obstetrics and Gynaecology Unit, University of Bari "Aldo Moro", 70124 Bari, Italy.

[2]Interdisciplinar Department of Medicine, University of Bari, 70124 Bari, Italy.

[3]Department of Medical and Surgical Sciences, University of Foggia, Foggia, Italy.

Received: May 22, 2024

Accepted: Jun 19, 2024

Published Online: Jun 26, 2024

Journal: Annals of Surgical Case Reports & Images

Online edition: https://annscri.org

Cite this article: Schönauer LM, Cerbone M, Bentivoglio M, de Nicolo M, Matteo M, et al. Herlyn-Werner-Wunderlich associated hematometra mimicking endometrioid neoformation: A rare case report. Ann Surg Case Rep Images. 2024; 1(4): 1033.

Introduction

In this case report we described a Mullerian anomaly associated with renal agenesia in a syndromic framework of Herlyn-Werner-Wunderlich syndrome. We underline the need of multidisciplinary counselling for those complex cases where the genetician, gynecologist, urologist should work together. The uniqueness of our case lies in the presence of a slightly symptomatic hematometra measuring 11 cm by 5 cm. This condition is noteworthy because it was initially misdiagnosed as an endometrial cyst on an MRI. The initial misdiagnosis highlights the challenge and importance of accurate imaging and diagnosis in such cases. Despite the hematometra's significant size, the patient exhibited only mild symptoms, adding another layer of complexity to the diagnosis and subsequent treatment plan.

Case report

We report the case of a 15-years-old woman who presented to our emergency room compelling for abdominal pain with a location in lower right iliac fossa. Her menarche was at age of 13; her menstrual period was reported irregular with oligo\ amenorrhoic menstruations with dismenorrea. She had congenital right renal agenesia and no family history for neoplastic pathologies. An abdominal ultrasound executed in ER revealed a dysomogeneous pelvic mass without vascular sign on the right side, near the adnexal area, suspect for endometirosic cyst. Near the cyst it was the evidence of a tubular neoformation of about 11 cm x 5 cm, suspect for sactosalpinx. No free fluid in the Douglas. The patient was hospitalized and subjected to full blood assessment, EKC, determination of oncomarkers.

The oncomarkers AFP, CEA, CA15.3, CA19.9 were negative with a slight augmentation of CA125 of 36,7 U/mL (Cutoff: 0-35 U/mL). Blood test and EKG was normal. Abdominal MRI showed a pelvic, paramedian right abdomino-pelvic mass of about 18 cm, with hourglass shape and dishomogeneous contrast enhancement, suspected for endometrioma, that compressed and dislocated the uterus and the sigmoid ansa to the left (Figure 1). The patient was subjected to hysteroscopy. Underwent general anestesia the patient went in the surgical room for the intervention. We performed a hysteroscopy without speculum with a 3.5 mm minihysteroscope (Versascope, Gynecare, Ethicon, Sommerville, NJ, U.S.A.) with saline solution as a distending medium. The vaginal space appeared narrowed because of the bulging hemivagina; we highlithed the presence of a right uterine cervix on the right and a vaginal septum on the left. The vaginal septum was punctured under transabdominal ultrasound guidance, but no material was drained. We converted the surgical intervention via laparotomy, revealing the presence of a uterus didelphys with the right uterus increased in volume and consistency compatible with hematometra and normal left uterus (Figure 2). The distal portion of the right salpinx showed multiple endometriotic implants. She had one vagina and a second cervix on the right side not connecting with the uterine cavity for the presence of a septum. She had a vaginal plastic surgery and hematometra drainage. This Mullerian anomaly associated with congenital unilateral renal agenesis suggests the presence of an Herlyn-Werner-Wunderlich syndrome. The post-operative period was uneventful and the hospital discharge was in III post-operative days. The hystologic exam was negative for neoplasy.

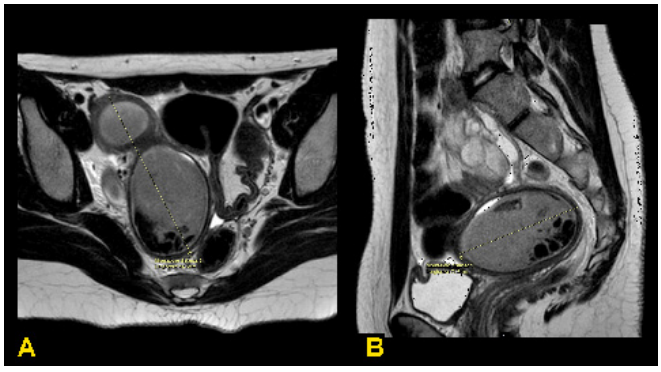

Figure 1: Abdominal MRI scan showing a pelvic mass reported to an endometrioma; **A:** transverse section **B:** sagittal section.

The abdominal MRI revealed a significant pelvic mass, situated in the paramedian right abdomino-pelvic region, measuring approximately 18 cm. This mass exhibited an hourglass shape and heterogeneous contrast enhancement. It was initially suspected to be an endometrioma. The size and location of the mass resulted in considerable compression and displacement of both the uterus and the sigmoid colon to the left.

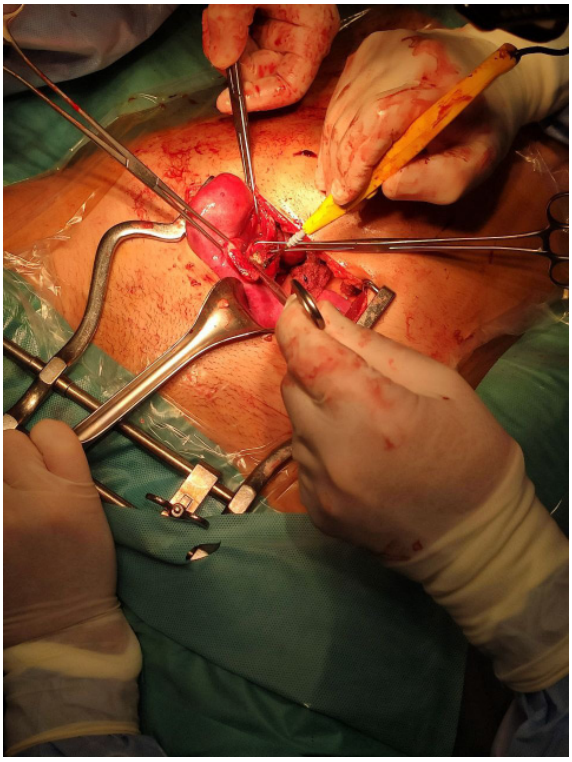

Figure 2: After hysterotomy, cutting of the uterine septum.

The right uterus was markedly enlarged and exhibited increased firmness, findings that were indicative of a hematometra. This notable enlargement and alteration in consistency suggested the accumulation of blood within the uterine cavity, a condition often associated with pain and other symptoms. In stark contrast, the left uterus maintained a normal appearance. It was of typical size and consistency, showing no signs of abnormality or disease. This clear distinction between the two uteri highlighted the asymmetrical nature of the condition and underscored the complexity of the patient's anatomical and pathological presentation.

Table 1: Main genes implicated in congenital syndromes involved in mullerian anomalies (Modified and adapted from [16]).

Syndrome	Inheritation	Etiology	Reproductive Anomaly	Other Findings
Acro-renal mandibular	AR	/	DU	Diaphragmatic hernia, ,
Antley-Bixler	AD	FGFR2 POR (AR)	VA	Choanal atresia,
Apert	AD	FGFR2	VA, BU	Cardiac disease
Cloacal exstrophy	/	/	Incomplete mullerian fusion	
Female pseudohermaphroditism with renal and gastrointestinal anomalies	/	/	DU Genital ambiguity, UA	KA
Female pseudohermaphroditism, renal and gastrointestinal anomalies	/	/	UA, Genital ambiguity,	KA, Gastrointestinal anomalies,
Fraser	AR	FRAS1, FREM2, GRIP1	VA, BU	KA, Mental retardation
Meckel	AR	MSK1, TMEM216, TMEM67, CEP290, RPGRIP1L, CC2D2A	BU Male pseudohermaphroditism	Dysplastic polycystic kidneys, encephalocele

Mosaic trisomy 7	/	Chromosomal	UA	Cystic kidneys,
MURCS association	/	/	VA, UA	KA
Pallister Hall	AD	GLI3	VA	cardiac defects
Roberts	AR	ESCO2	BU, UA , VA	Cardiac defects
Roberts	AR	ESCO2	UA and VA	Tetraphocomelia,, cardiac defects
Rüdiger	AR	/	BU	ureteral stenosis, mental retardation
Urogenital adysplasia	/	/	Unicornuate or BU	KA

Note: AD: Autosomic Dominant; AR: Autosomic Recessive; VA: Vaginal Atresia; BU: Bicornuate Uterus; UA: Uterus Agenesia; DU: Didelphys Uterus; KA: Kidney Anomalies; KA: kidney Agenesia.

Discussion

Congenital Uterine Anomalies (CUA) result from abnormal formation, fusion or resorption of the Müllerian ducts during fetal life [1], that are usually detected incidentally during fertility investigations: CUA's could be asymptomatic and most women with uterine anomalies could have a normal reproductive outcome; however some women may experience adverse reproductive outcome with an increased rate of miscarriage, preterm delivery and other adverse fetal outcomes [2-8]. In the general population the incidence of CUA is about 7%, with a prevalence of arcuate (68%) and septate uterus (27%) and a rarity of bicornuate (4%) and didelphys (0.4 %) uterus. The incidence of CUE in infertile population is almost identical to general population, with a predominance of septate uterus (46%) and arcuate uterus (25%). Patients diagnosed with multiple miscarriages have a higher prevalence of CUA, accounting for about 17% of cases, with a prevalence of arcuate uterus (65%). [9-12] CUA may be associated with congenital renal anomalies due to a close embryologic relation between the development of the urinary and reproductive organs [13]: Evaluation of the genital tract is recommended for women with major urologic anomalies. Congenital uterine anomalies are not uncommon: reported population prevalence rates in individual studies varying between 0.06% and 38%, and the observed wide variation is possibly due to the assessment of different study populations and the use of different diagnostic techniques [14]. Chan et al. conducted a systematic review of studies evaluating the prevalence of congenital uterine anomalies in the unselected population and in women with a history of infertility, including those undergoing IVF treatment, miscarriage, infertility and recurrent miscarriage combined, and preterm delivery. This review evaluated that the prevalence of uterine anomalies diagnosed by optimal tests was 5.5% in an unselected population, 8% in infertile women, 13.3% in those with miscarriage and highest at 24.5% in infertile women who also had a history of miscarriage [9]. There are many classifications of CUAs (Table 2): The first of these reported by Cruveilher, Foerster and von Rokitansky in the mid-19th century, then the classification introduced by Buttram and Gibbons in 1979 that was later revised by the American Fertility Society (AFS), now known as the American Society of Reproductive Medicine (ASRM) [15]. The anomalies were classified as: hypoplasia/agenesis, unicornuate, didelphys, bicornuate, septate, arcuate and Diethylstilboestrol (DES) drug-related. However this classification included only uterine anomalies with the exclusion of cervical and vaginal anomalies, did not classify combined or complex anomalies and the arcuate uterus being included as a separate class. The European Society of Human Reproduction and Embryology (ESHRE) and the European Society for Gynecological Endoscopy (ESGE) developed a new updated classification system through a structured Delphi procedure [7]. Uterine anomalies are classified into seven main types: U0, normal uterus; U1, dysmorphic uterus (infantile or T-shaped); U2, septate uterus; U3, bicorporeal uterus (partial and complete—bicornuate and didelphys); U4, hemi uterus (unicornuate based on AFS); U5, aplastic uterus; U6, for unclassified cases. Combined laparoscopy and hysteroscopy in considered the gold standard test among the main diagnostic tools, which also includes ultrasonography, hysterosalpingogram, sonohysterogram and MRI. Hysterosalpingogram, a common instrument for fertility investigation, can evaluate the uterine cavity but can't study the external uterine contour and can't differentiate between bicornuate and septate uteri. 2D transvaginal ultrasound is minimally invasive and a less expensive way to study uterine morphology, however 3D transvaginal ultrasound is considered the less invasive gold standard tool for the study of uterine anomalies: three orthogonal planes can be viewed in different modes to study the external and internal uterus contours. The diagnostic accuracy of 3D ultrasound is reported as 97.6% with sensitivity and specificity of 98.3% and 99.4% respectively [7].

Conclusion

Müllerian anomalies, though not uncommon, can present significant diagnostic challenges due to their diverse manifestations. In this paper, we explored a particularly rare syndrome that necessitated a complex and multi-disciplinary approach, combining both surgical and hysteroscopic techniques. The intricacies of this case underscore the importance of a comprehensive diagnostic and therapeutic strategy to address such unique and multifaceted conditions effectively. Our discussion detailed the necessity for a collaborative effort among various medical specialties, including gynecology, radiology, and surgery. This multidisciplinary team approach is essential not only for accurate diagnosis but also for the development of a tailored treatment plan that meets the specific needs and expectations of the patient. By involving specialists from different fields, we can ensure a more thorough understanding of the condition and provide a higher standard of care. Moreover, this case emphasizes the importance of considering the patient's individual circumstances and goals in the treatment plan. The involvement of a diverse medical team allows for a more holistic view, integrating different perspectives and expertise to achieve the best possible outcomes. Therefore, we advocate for the establishment of specialized centers with dedicated teams to manage complex Müllerian anomalies, as this approach is crucial for optimizing patient care and advancing our understanding of these rare conditions.

Declarations

Author contributions: All authors approved the final version of the manuscript.

Funding: This research received no external funding.

Study registration: Not applicable

Disclosure of interests: The authors declare no conflicts of interests

Ethical approval: Ethics committee approval was not necessary since the study was a summary of data and outcomes of routine management (without direct intervention) and not an experimental protocol. We ensured the complete anonimacy of the patients.

Informed consent statement: Written informed consent was obtained from the patient to publish this paper.

Data availability statement: All data are reported in the text. The data that support the findings of this study are available on the main medical databases.

References

1. Pfeifer SM, Attaran M, Goldstein J, Lindheim SR, Petrozza JC, et al. ASRM müllerian anomalies classification 2021. Fertil Steril. 2021; 116(5): 1238-52.

2. Bortoletto P, Romanski PA, Pfeifer SM. Müllerian Anomalies: Presentation, Diagnosis, and Counseling. Obstet Gynecol. 2024; 143(3): 369-77.

3. Ludwin A, Pfeifer SM. Reproductive surgery for müllerian anomalies: A review of progress in the last decade. Fertil Steril. 2019; 112(3): 408-16.

4. Green LK, Harris RE. Uterine anomalies. Frequency of diagnosis and associated obstetric complications. Obstet Gynecol. 1976; 47(4): 427-9.

5. Rock JA, Schlaff WD. The obstetric consequences of uterovaginal anomalies. Fertil Steril. 1985; 43(5): 681-92.

6. Raga F, Bauset C, Remohi J, Bonilla-Musoles F, Simón C, et al. Reproductive impact of congenital Müllerian anomalies. Hum Reprod. 1997; 12(10): 2277-81.

7. Grimbizis GF, Di Spiezio Sardo A, Saravelos SH, Gordts S, Exacoustos C, et al. The Thessaloniki ESHRE/ESGE consensus on diagnosis of female genital anomalies. Gynecol Surg. 2016; 13: 1-16.

8. Tomazevic T, Ban-Frangez H, Ribic-Pucelj M, Premru-Srsen T, Verdenik I. Small uterine septum is an important risk variable for preterm birth. Eur J Obstet Gynecol Reprod Biol. 2007; 135(2): 154-7.

9. Saravelos SH, Cocksedge KA, Li TC. Prevalence and diagnosis of congenital uterine anomalies in women with reproductive failure: a critical appraisal. Hum Reprod Update. 2008; 14(5): 415-29.

10. Prasannan L, Rekawek P, Kinzler WL, Richmond DA, Chavez MR. Assessing müllerian anomalies in early pregnancy utilizing advanced 3-dimensional ultrasound technology. Am J Obstet Gynecol. 2024; S0002-9378(24)00529-5.

11. Fedele L, Vercellini P, Marchini M, Ricciardiello O, Candiani GB. Communicating uteri: Description and classification of a new type. Int J Fertil. 1988; 33(3): 168-72.

12. Grimbizis GF, Camus M, Tarlatzis BC, Bontis JN, Devroey P. Clinical implications of uterine malformations and hysteroscopic treatment results. Hum Reprod Update. 2001; 7(2): 161-74.

13. Heinonen PK. Distribution of female genital tract anomalies in two classifications. Eur J Obstet Gynecol Reprod Biol. 2016; 206: 141-6.

14. Chan YY, Jayaprakasan K, Zamora J, Thornton JG, Raine-Fenning N, et al. The prevalence of congenital uterine anomalies in unselected and high-risk populations: a systematic review. Hum Reprod Update. 2011; 17(6): 761-71.

15. The American Fertility Society classifications of adnexal adhesions, distal tubal occlusion, tubal occlusion secondary to tubal ligation, tubal pregnancies, müllerian anomalies and intrauterine adhesions. Fertil Steril. 1988; 49(6): 944-55.

16. Connell M, Owen C, Segars J. Genetic Syndromes and Genes Involved in the Development of the Female Reproductive Tract: A Possible Role for Gene Therapy. J Genet Syndr Gene Ther. 2013; 4: 127.