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REVISITING MULLERIAN ANOMALIES: A PICTORIAL REVIEW

Radio-Diagnosis

Dr. Aman Gangwani	PG JR-3, Dept. Of Radiodiagnosis JNUIMSRC, Jaipur.
Dr. Usha Jaipal	Prof. And HOD, Dept. Of Radiodiagnosis, JNUIMSRC, Jaipur.
Dr. Rajeev Mudkavi	Professor, Dept Of Radiodiagnosis, JNUIMSRC, Jaipur.
Dr. Naveen Meena	Professor, Dept Of Radiodiagnosis, JNUIMSRC, Jaipur.

ABSTRACT

Utero-vaginal anomalies result from defects in the development or fusion of the Müllerian ducts. These anomalies are associated with a high incidence of altered fertility and obstetric problems. Uterovaginal anomalies are generally classified into dysgenesis, vertical or lateral fusion anomalies and abnormal configuration. Sequential analysis of USG followed by MR images enables accurate morphological and anatomical demonstration of parameters like: uterine size, external fundal contour, intercornual distance, and presence of uterine or vaginal septa, thereby assisting in a proper treatment approach. Furthermore, other associated/isolated pelvic lesions or renal anomalies can also be detected. MRI further helps in the evaluation of obstructive uterovaginal anomalies and determining the site of obstruction and surgical approach. Imaging findings of uterovaginal anomalies, which presented as abdominal pain and altered menstrual cycles, are shown. **Objective:** Radiological evaluation of mullerian anomalies with non-specific and overlapping clinical presentation sensitization of the relevant specialities involved for improving diagnosis and preventing further complications. **Materials And Methods:** Relevant radiological investigations were carried out on opd and ipd patients in the department of radiodiagnosis at jaipur national university institute for medical sciences and research centre, jaipur. Usg – initial modality of choice and later on patients were referred for MRI, where relevant sequences were taken.

KEYWORDS

Herlyn-Werner-Wunderlich syndrome, Mullerian anomalies, obstructed hemi vagina, ipsilateral renal agenesis, uterus didelphys, ACUM (accessory and cavitated uterine mass), Bicornuate Unicollis Uterus, arcuate uterus.

INTRODUCTION:

Mullerian anomalies have a significant impact on the reproductive health of female patients. For patients presenting with both infertility and recurrent pregnancy loss, the incidence of Mullerian anomalies may go as high as 25%. Congenital anomalies of the uterus are often just one part of a complex set of anomalies also involving the cervix, vagina and the urinary tract. Recognition of typical features of these anomalies is necessary for timely diagnosis and management of the patient, as these can have overlapping clinical features like pelvic pain, abnormal menstruation (dysmenorrhea/oligomenorrhea/amenorrhea), failure to conceive / if the patient conceives, recurrent pregnancy loss, pelvic mass, which can create a diagnostic dilemma. Once a Mullerian anomaly has been identified, further evaluation should be performed for any associated urinary tract anomalies.

In patients presenting with infertility and seeking the use of assisted reproductive technologies(ART), recognition of Mullerian anomalies may affect reproductive success, especially in cases that have an incomplete / clinically occult septum that divides the cervix.

USG is usually the first choice of investigation for evaluating the pelvic organs, and later, the patient can be referred for MRI, which further helps in visualising the external uterine fundal contour and internal indentation of the endometrial cavity. These two crucial morphological characteristics are the keys to diagnosing congenital anomalies.

Arcuate Uterus

This entity is a mild form of Mullerian duct anomaly and is characterised by a broad base smooth indentation on the fundal endometrial canal and a normal external fundal contour.

On imaging, the normal convex external uterine contour is maintained, and there is a broad-based smooth indentation of the endometrial cavity.

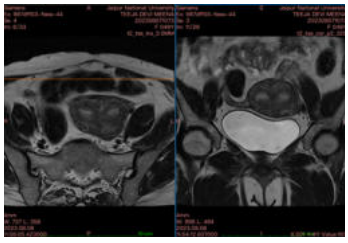

Figure A.

A case of a 48-year-old female with an incidental finding of an arcuate uterus. There is evidence of central myometrial indentation in the fundal region of the uterus with normal external uterine contour.

Septate Uterus

Accounting for about 55% of the cases, septate uterus is the most common Mullerian duct anomaly. Sensitisation towards this entity is crucial as it has a strong association with recurrent miscarriages and can be suspected in patients who come with a history of mid-trimester pregnancy loss. The usual diagnostic dilemma in patients suspected to have a Mullerian duct anomaly is differentiating between a bicornuate and a septate uterus. In cases of a septate uterus, a normal convex external fundal contour will be seen.

The origin of the septum is from the midline of the uterine fundus, and it results from partial failure or complete reabsorption of the utero-vaginal septum. There can be a complete or a partial septum where a complete septum may extend to the external Os and in some cases even upto the vagina. The length and the composition of the septum may vary, consisting of different proportions of myometrium and fibrous tissue.

On USG, the myometrium may be seen interrupted by a septum at the fundus, while on MRI, the key differentiating feature between a septate and bicornuate uterus is the external fundal contour. A clear fundal cleft is highly indicative of fusion anomalies like bicornuate uterus rather than reabsorption anomalies like septate or arcuate uterus.

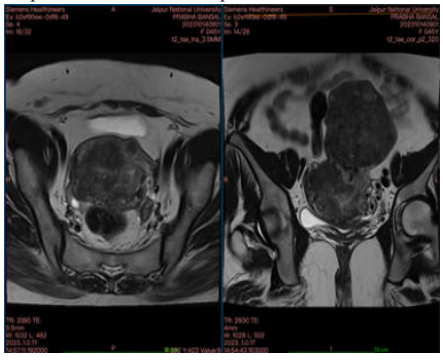

Figure B.

A case of a 45-year-old female with septate morphology of the uterus and a large subserosal fibroid.

It is crucial to identify the composition of the septum on MRI (myometrium has high signal on T2-weighted images while the fibrous septum has low signal), as it changes the surgical approach, a muscular septum may require a transabdominal surgical approach, while the fibrous septum can be treated by a less invasive hysteroscopic septoplasty.

Bicornuate Uterus

This anomaly is a result of the partial or incomplete fusion of the Müllerian ducts. The characteristic feature of a bicornuate uterus is the presence of a cleft of more than 1 cm on the uterine fundus, which is similar to that of didelphys. The duplication of the endometrial cavity may or may not be associated with duplication of the cervix (Bicollis/Unicollis). In case of presence of a longitudinal vaginal septum this entity may be indistinguishable from uterus didelphys.

On USG, two divergent uterine horns with separate endometrial cavities may be demonstrated due to echogenicity of the endometrium (depending on the phase of the menstrual cycle), while on MRI, normal zonal anatomy of the uterine horns is well visualised. In the absence of vaginal duplication, the two separate cervices (which may give the appearance of "eyes of owl") are noted in patients with bicornuate bicollis uterus.

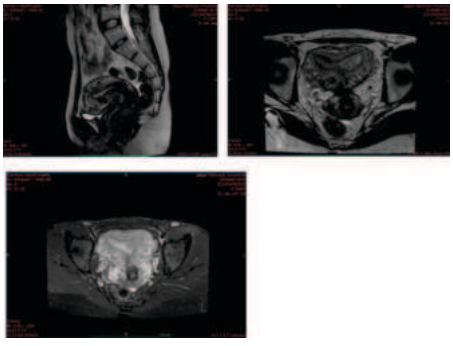

Figure C.

A 28-year-old married female presented with a history of continuous bleeding for 2 months (AUB). K/c/o hypothyroidism on medication.

Two widely divergent uterine horns are noted with inter-cornual distance of ~5.5 cm and single cervix and vagina suggestive of bicornuate Unicollis uterus. CLASS U3A/C0/V0

Uterus Didelphys

This Mullerian anomaly is a result of complete failure of Mullerian duct fusion, where each duct develops completely with duplication of uterine horns, cervix and proximal vagina.

The duplicated proximal vagina can be associated with a transverse septum with resultant ipsilateral obstruction and hematometra/ hematocolpos. However, in the absence of vaginal obstruction uterus didelphys is asymptomatic.

Patients with obstructed hemivagina can present with dysmenorrhea, infections and adhesions as a result of retrograde menstrual flow. Obstructed uterus didelphys is usually associated with ipsilateral renal agenesis.

On ultrasound, the differentiation of reabsorption anomalies (arcuate & septate) from fusion anomalies (bicornuate & didelphys) is done based on uterine fundal cleft, which can be demonstrated on true coronal images. In case of uterus didelphys two widely divergent uterine horns are present with non-communicating endometrial cavities along with two cervices and duplicated upper vaginas. In patients where duplication of vagina is not apparent differentiating between uterus didelphys and bicornuate bicollis uterus becomes difficult.

On MRI, similar to USG, two widely divergent uterine horns and separate cervices can be demonstrated. "A fundal cleft >1 cm has been reported to be 100% sensitive and specific in differentiating fusion anomalies from reabsorption anomalies".

In cases of uterus didelphys, zonal anatomy and endometrial to myometrial ratios are generally normal. One of the uterine horns will be markedly distended from blood products in cases of the presence of a hemi-vaginal septum, which will be seen as a high signal intensity on T1-weighted images.

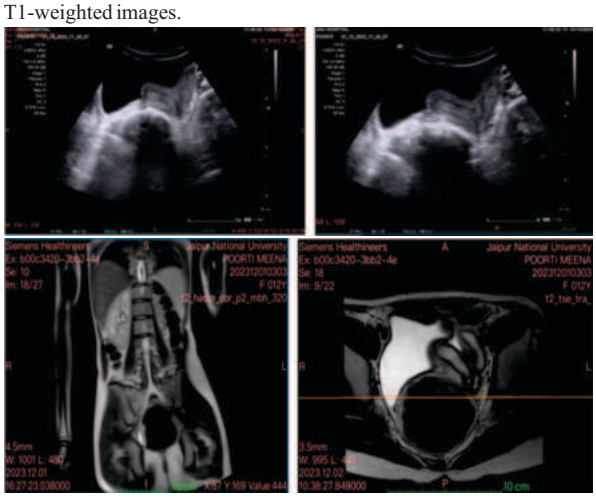

Figure D.

A 12-year-old female presented with a history of continuous bleeding per vaginum for 1 month. Menarche – 1 month. Thyroid profile – elevated TSH levels.

Two divergent, distinct uterine horns with a cleft in between are noted. Two separate complete endometria were noted - uterine didelphys. The left kidney was not visualised in the left renal fossa.

Two widely divergent horns are seen with two separate cervices – suggestive of uterus didelphys (complete Bicorporeal uterus with double cervix). No evidence of Hematocolpos noted at present.

Herlyn-Werner-Wunderlich Syndrome / Ohvira – Obstructed Hemivagina With Ipsilateral Renal Anomalies

OHVIRA is a rare complex Mullerian anomaly; it includes the triad of uterus didelphys (duplication of uterus), obstructed hemivagina and ipsilateral renal agenesis.

OHVIRA syndrome is classified under class III according to the ASRM classification and class U3b C2 V2 as per ESHRE classification. OHVIRA variants may at times be associated with a bicornuate or septate uterus. As per a retrospective study by Zhu et al. OHVIRA syndrome is classified into two classes based on whether the obstruction of the hemivagina is complete or incomplete.

For the diagnosis of OHVIRA multimodal approach is required, which includes a detailed history of the patient, clinical examination and imaging studies. As per the American Academy of Paediatrics, routine examination of genitalia in the adolescent age group is recommended in case of genitourinary symptoms. Imaging modalities such as USG and MRI are established in aiding diagnosis, where a 3D ultrasound further increases the sensitivity and specificity of diagnosis and can be considered an alternative to MRI. Various difficulties may arise in diagnosing the condition due to small size of the uterus, inactive endometrium dilated vagina in peripubertal females.

MRI provides detailed information about the uterine anatomy and, therefore, is considered the gold standard investigation for the diagnosis of this condition.

Adequate and timely treatment of obstructed anomalies is crucial to prevent chronic complications and negative psychological impact. Awareness and a high index of suspicion for such conditions would help in early diagnosis and relief of obstructive symptoms with preservation of reproductive function. Though a complex and rare anomaly treatment of OHVIRA is relatively simple, either resection or excision of vaginal septum.

Other causes of outflow obstruction which can be considered as differential diagnosis include imperforate hymen, transverse/ longitudinal vaginal septum and atresia of cervix.

Follow-up of such patients is essential due to the associated risk of adhesions, stenosis and endometriosis.

Figure E.

A 17-year-old unmarried female presented with a history of pain and heaviness in the lower abdomen cyclically associated with the onset of menstruation. No history of abnormal vaginal discharge or fever. She had well-developed secondary sexual characteristics appropriate for her age, including breast, axillary and pubic hair.

USG: UTERUS: Two diverging endometria noted in the upper segment with deep indentation/cleft of more than 1 cm with obtuse angle divergence along the fundus and upper segment – suggestive of Bicornuate uterus (Didelphys).

A well-defined hypoechoic collection is noted extending from the left lower endometrial cavity and along the vagina with Fluid – Fluid levels measuring approx. 98 x 63 x 51 mm likely suggestive of Hematocolpos.

Left Kidney: Not visualised in the left renal fossa, suggestive of an absent kidney.

MRI: Two Widely Divergent Uterine Horns Are Seen With Two Separate Cervices And Longitudinally Separate Upper Vagina – Suggestive Of Uterus Didelphys (complete Bicorporeal Uterus With Double Cervix.)

The left hemivagina is obstructed with resultant hematocolpos appearing t1 iso to hyperintense. The left kidney is not visualised in the left renal fossa.

ACUM - Accessory And Cavitated Uterine Mass

ACUM, a relatively rare uterine anomaly, is typically characterised by the presence of a non-communicating accessory uterine cavity with no other obstructive congenital uterine anomalies.

However, the exact pathogenesis of this entity is not known; it is believed to be a newly recognised Mullerian anomaly with developmentally displaced Mullerian tissue, while some suggest it is a form of metaplasia. Various other terms have been used for ACUMs – uterine-like mass, adenomyotic cysts, juvenile cystic adenomyosis and myometrial cysts.

At present, ACUM is not classified in the ESHRE, ESGE and rASRM classification of uterine anomalies and is still possibly an underdiagnosed clinical entity.

Stefan Timmerman et al. proposed a definition modified from Naftalin et al.: "a cavitated lesion surrounded by myometrial mantle, in continuity with the antero-lateral uterine wall and located beneath the insertion of the round ligament and the interstitial portion of the fallopian tubes". For distinguishing ACUM from other obstructive uterine anomalies, a normal uterine cavity should be visualised.

Various management approaches have been reported, with surgical excision being performed in most of the cases and cornual excision and total hysterectomy in some.

Recent literature shows the use of USG-guided sclerotherapy with the rationale of destruction of the functional endometrium and prevention of accumulation of menstrual blood.

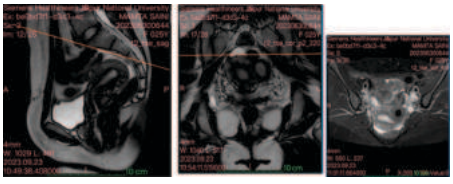

Figure F.

A 25-year-old unmarried female presented with severe pain during menstruation since menarche, which had aggravated a few months before the hospital visit. Her menstrual cycles were irregular. There was no history suggestive of pelvic inflammatory disease. Her general physical and per abdominal examination was normal.

The uterus is midline and appears normal in size. The cervix and vagina are single and appear normal. A blind-ended cavity is seen in the left lateral uterine wall under the round ligament with endometrial lining. Blood degradation products are noted in the cavity. Bilateral uterine cornuae appear normal.

MRKH - Mayer-rokitansky-kuster-hauser Syndrome / Mullerian Agenesis

Also known as Rokitansky syndrome is a caused due to interrupted embryonic development of the paramesonephric ducts resulting in hypoplasia of the uterus and the upper 2/3rd of the vagina. Patients usually present with primary amenorrhea in the setting of normal pubertal and sexual development (due to normal ovarian function).

Timely diagnosis with appropriate clinical and psychological input is essential, as this syndrome has devastating effects on the fertility and sexual life of young women.

The presence of rudimentary Mullerian structures may result in diagnostic uncertainty due to a common misconception that patients with MRKH have no uterine development. Ovaries in such patients are normal but can be ectopic.

Females with MRKH usually never menstruate or carry a pregnancy, however, ovum donation / surrogate pregnancy might be considered as an option. Furthermore, the absence of vagina necessitates the need of treatment for allowing penetrative intercourse.

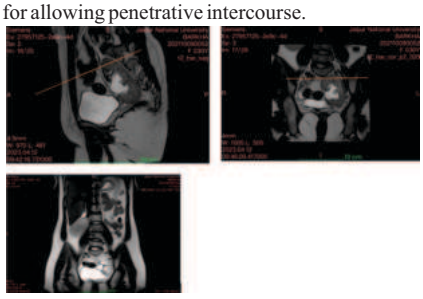

Figure G.

A case of a 30-year-old female with primary amenorrhea.

Uterus and cervix are not visualised with non-visualisation of upper part of vagina, left kidney is ectopically placed in the pelvic cavity on left side with hila facing anteriorly and dilated PCS, Right kidney lies in right renal fossa and appears normal.

Absence of uterus, cervix and upper part of vagina and normal bilateral ovaries consistent with mayer-rokitansky-kuster-hauser syndrome / mullerian agenesis

CONCLUSION:

Herlyn-Werner-Wunderlich syndrome, aka OHVIRA syndrome (obstructed hemivagina with ipsilateral renal anomalies), ACUM (accessory and cavitated uterine mass), bicornuate uterus are rare congenital anomalies of the female genito-urinary tract, presenting with symptoms like dysmenorrhea, pelvic pain, pelvic mass, which may be persistent and resistant to initial treatment and can further complicate to endometriosis, infertility and recurrent miscarriages if are misdiagnosed/are detected at a later stage / remain undetected due to non-sensitization to such entities. Therefore, there is a need to sensitise the gynecologists, Paediatricians and radiologists to these rare entities to clear out the diagnostic dilemma associated with these entities for timely diagnosis, intervention and prevention of future complications.

Strengths And Limitations:

- **Strengths:** excellent non-invasive imaging with visualisation of detailed anatomy/anomalies, with better patient compliance and opportunity for earlier corrective approach.
- **Limitations:** The scope is limited and may require further exploration through invasive procedures, such as laparoscopy or laparotomy.

REFERENCES

1. RTowbin RB, Schaefer CM, Li Y, Towbin AJ. Obstructed Hemivagina with Ipsilateral Renal Anomalies.
2. Patil B, Nagaraju RM. Uterine didelphys with obstructed hemivagina and ipsilateral renal anomaly-OHVIRA syndrome: a rare congenital anomaly. International Journal of Reproduction, Contraception, Obstetrics and Gynecology. 2015 Jun 1;4(3):889-93.
3. Sharma R, Mishra P, Seth S, Agarwal N. OHVIRA syndrome—diagnostic dilemmas and review of literature. Journal of South Asian Federation of Obstetrics and Gynaecology. 2020 Nov;12(6):422.
4. Del Vescovo R, Battisti S, Di Paola V, Piccolo CL, Cazzato RL, Sansoni I, Grasso RF, Zobel BB. Herlyn-Werner-Wunderlich syndrome: MRI findings, radiological guide (two cases and literature review), and differential diagnosis. BMC Medical Imaging. 2012 Dec;12:1-0.
5. Sugi MD, Penna R, Jha P, Põder L, Behr SC, Courtier J, Mok-Lin E, Rabban JT, Choi HH. Müllerian duct anomalies: role in fertility and pregnancy. Radiographics. 2021 Oct;41(6):1857-75.
6. Bajaj SK, Misra R, Thukral BB, Gupta R. OHVIRA: Uterus didelphys, blind hemivagina and ipsilateral renal agenesis: Advantage MRI. Journal of human reproductive sciences. 2012 Jan;5(1):67.
7. Herlyn-Werner-Wunderlich syndrome - OHVIRA syndrome: a rare congenital anomaly. Section Genital (female) imaging .Case Type Clinical Cases. Authors Shaista Shoukat, Warda Sattar, Kausar Illahi Bux.. Jinnah Post Graduate Medical College Karachi; Rafique shaed road Karachi 75510 Karachi, Pakistan; https://www.eurorad.org/case/13438 10.1594/EURORAD/CASE.13438. 1563-4086
8. Hall-Craggs MA, Williams CE, Pattison SH, Kirkham AP, Creighton SM. Mayer-Rokitansky-Kuster-Hauser syndrome: diagnosis with MR imaging. Radiology. 2013 Dec;269(3):787-92.
9. Bajaj SK, Misra R, Thukral BB, Gupta R. OHVIRA: Uterus didelphys, blind hemivagina and ipsilateral renal agenesis: Advantage MRI. J Hum Reprod Sci. 2012 Jan;5(1):67-70. doi: 10.4103/0974-1208.97811. PMID: 22870020; PMCID: PMC3409925.
10. Sharma R, Mishra P, Seth S, et al. OHVIRA Syndrome—Diagnostic Dilemmas and Review of Literature. J South Asian Feder Obst Gynae 2020;12(6):421–426.
11. Timmerman S, Stubbe L, Van den Bosch T, Van Schoubroeck D, Tellum T, Froyman W. Accessory cavitated uterine malformation (ACUM): A scoping review. Acta Obstetricia et Gynecologica Scandinavica. 2024 Jun;103(6):1036-45.