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PRIMARY AMENORRHEA WITH APLASTIC UTERUS, BILATERAL NON-COMMUNICATING RUDIMENTARY HORNS WITH HEMATOMETRA IN LEFT HORN AND CERVICOVAGINAL APLASIA (U5A C4 V4) - A RAREST CASE REPORT

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Keywords:

Congenital uterine malformations, *Mullerian agenesis*, Primary amenorrhea, aplastic uterus, Cervicovaginal aplasia, Vaginoplasty

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ABSTRACT: Congenital uterine malformations account for 5.5 % of unselected population, while mullerian agenesis has an incidence of 1 in 4500-5000 females. Mullerian agenesis occurs due to embryological underdevelopment of mullerian duct with resultant agenesis or atresia of the vagina, uterus or both. We report a rare case of 22 year old female with primary amenorrhea with aplastic uterus with bilateral rudimentary horns, hematometra in left rudimentary horn and Cervicovaginal aplasia. She came to our OPD with history of primary amenorrhea and vaginal discharge for 15 days. General physical examination revealed well developed secondary sexual characteristics and normal breast development. Axillary and pubic hair development was normal. A small dimple of 1 cm was noted at the site of Vagina. Uterus was felt like a small hypoplastic nodule on per rectal examination. Laparoscopy revealed bilateral rudimentary horns , combined agenesis of uterus , cervix and vagina. Resection of Bilateral rudimentary horns with hematometra and bilateral salpingectomy was done followed by Mc Indioe Vaginoplasty on follow up visit 3 months before her wedding. 3D Ultrasonography and MRI are gold standard for diagnosing Mullerian agenesis, but using laparoscopy aids in arriving at an accurate diagnosis. Patients should be provided with mental health support for better social, marital life and gestational surrogacy as the fertility option.

INTRODUCTION: Congenital uterine malformations account for 5.5 % of unselected population while mullerian agenesis has an incidence of 1 in 4500-5000 females [1]. Mullerian agenesis occurs due to embryological underdevelopment of mullerian duct with resultant agenesis or atresia of the vagina, uterus or both. Müllerian aplasia, Mayer-Rokitansky-Küster-Hauser syndrome, or vaginal agenesis are the synonyms for mullerian Agenesis.

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Evaluation of a patient with primary amenorrhea should include a detailed history, physical examination including tanners staging and complete laboratory work up. On evaluating women with Mullerian abnormalities vaginal canal is markedly shortened and may appear as a dimple below the urethra [2]. A single midline uterine remnant may be present or uterine horns (with or without an endometrial cavity) may exist.

The ovaries, as they have separate embryologic source, are typically normal in structure and function, though they may be found in atypical locations [3]. MRKH syndrome is frequently associated with malformation which are recently classified as typical (isolated utero vaginal aplasia / hypoplasia) and atypical (additional renal (27-29%) skeletal (8-32 %) and ovarian abnormalities) [4].

We report a rare case of 22-year-old female with primary amenorrhea with aplastic uterus with bilateral rudimentary horns, hematometra in left rudimentary horn and cervicovaginal aplasia.

Case Description: A 22-year-old unmarried female came to our OPD with a history of primary amenorrhea and vaginal discharge for 15 days. A general physical examination revealed well-developed secondary sexual characteristics and normal breast development. Axillary and pubic hair development was normal. A small dimple of 1 cm was noted at the site of Vagina. The distance between vaginal dimple and anus was 5 cm. Uterus was felt like a small hypoplastic nodule on per rectal examination. No history of cyclical abdominal pain or discharge from breast. Ultrasonography showed hypoplastic uterus with bilateral ovaries normal. Buccal smear showed presence of Barr body. Investigations revealed normal serum testosterone, thyroid profile and LH. Patient had elevated prolactin and DHEAS. MRI whole abdomen and pelvis **Fig. 1** revealed the absence of uterus and cervix suggestive of agenesis. Vagina was also hypoplastic and fairly shortened. Left fallopian tube was dilated measuring 13 mm and showed chronic blood products appearing hypointense on T1 W / T2 W. SPIR image with fluid levels on T2 weighted image was suggestive of hematosalpinx. Small endometrioma 9 mm X 8 mm in size seen in left ovary. Right ovary was normal size. A provisional diagnosis of combined agenesis of uterus, cervix and vagina, left hematosalpinx and left ovarian endometrioma after MRI report.

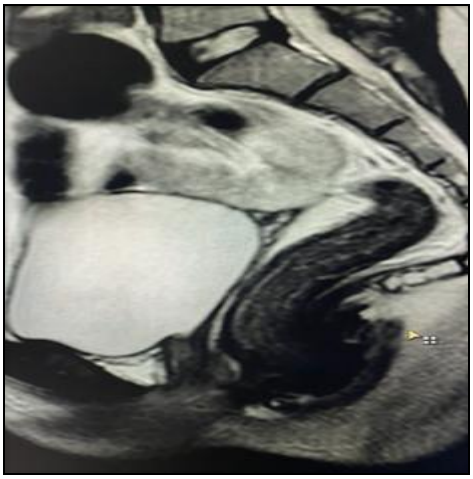

FIG. 1: MRI IMAGE SHOWING THE ABSENCE OF UTERINE TISSUE IN BETWEEN THE URINARY BLADDER AND RECTUM

The patient was taken up for diagnostic laparoscopy and proceeded **Fig. 2.**

Intraoperative findings:

- Bilateral rudimentary horns of uterus seen separated by 10 cm transverse band of 1 cm thickness.
- Left rudimentary horn of size 1.5x1 cm with hematometrawas seen. However bilateral fallopian tubes were normal.
- Corpus luteal cyst was seen in left ovary. Right ovary was normal.
- Combined agesis of uterus, cervix and vagina was noted.
- Resection of Bilateral rudimentary horns with hematometra and bilateral salpingectomy was done.

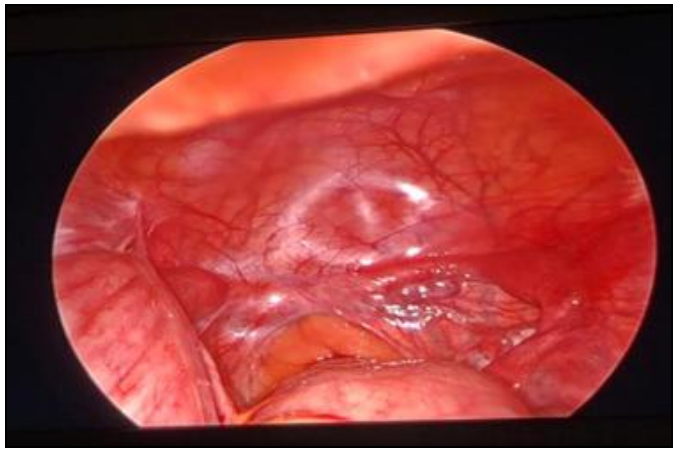

FIG. 2: LAPAROSCOPY IMAGE SHOWING RIGHT AND LEFT RUDIMENTARY HORNS WITH DEFICIENT UTERINE TISSUE IN BETWEEN

Diagnosis as per ESHRE classification U5a C4 V4 - Aplastic uterus with bilateral non-communicating rudimentary horns with hematometra in the left rudimentary horn with cervicovaginal aplasia was made [5]. The patient tolerated the procedure well and discharged on POD 5. The patient and attendants were counselled regarding her condition, management plan, future fertility options, associated risks, and the need for mental and social support. Mc Indioe Vaginoplasty was done on follow up visit 3 months prior to her wedding. Repeated dressing with vaginal mould was done. Patient was discharged 4 weeks after surgery. Psychosocial counselling and moral support were

part of the management throughout. Patient is on regular follow up with satisfied marital life.

DISCUSSION: Primary vaginal elongation by dilatation is the first line approach in most of the patients with mullerian agenesis as it is safer, patient controlled and more cost effective. Creation of neo vagina by vaginal dilatation is one recommended procedures. Vaginoplasty by different surgical methods seems to have the highest anatomical (99%) and functional (96 %) success rates [6]. If needed surgical procedures are often advised to patients three to six months prior to marriage [7]. Correction of the congenital anomaly along with psychosocial counselling remains the cornerstone for maximum efficacy of treatment. Encouragement to connect with peer support groups should be given. For Conception ART, IVF with gestational surrogates can give promising results for the patients with Mullerian anomalies. Adoption can be offered as an alternative option. Uterine transplantation in the near future seems to give promising results to provide motherhood to patients with combined uterine agenesis.

CONCLUSION: Most Mullerian anomalies remain undiagnosed due to the lack of clinical suspicion and absence of pathognomic clinical and radiological characteristics. All the cases with Mullerian agenesis present with primary amenorrhea as the cardinal symptom. 3D Ultrasonography and MRI are gold standard to diagnose Mullerian agenesis but use of laparoscopy aids in arriving at an accurate diagnosis [8]. Patients should be provided with mental health support for better social, marital life and gestational surrogacy as the fertility option.

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CONFLICTS OF INTEREST: None

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