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A Study of Congenital Abnormalities of Female Genital Tract: In A Tertiary Care Center

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ABSTRACT

Introduction: Developmental anomalies of female genital tract are not very common, incidence reported in the literature is0.2%–3.8% [1] Depending on the specific defect, a woman's obstetric and gynaecologic health may be adversely affected.

Aims and Objectives: Through the present study we aim to evaluate the various clinical presentations of congenital anomalies of female genital tract, appropriate management used for the respective cases to improve both obstetric and gynaecological outcome.

Methods: A prospective observational study was conducted at the Maharajah's Institute of Medical sciences, Obstetrics and Gynaecology department for a period of 3 years. Among more than 3000 patients reporting in the outpatient department within the above mentioned time period, 13 cases of female genital tract developmental defects were diagnosed through clinical history, physical examination, hormonal study, imaging-studies and hysterolaparoscopy. The optimum mode of management for each case was individualised and assessed in terms of the final outcome and is being followed-up.

Results: Present study shows prevalence of mullerian anomalies is 0.4 % at our tertiary care center. Main presenting symptoms were primary amenorrhoea and infertility. Most of the patients belonged to the adolescent age group.

Conclusion: Female genital tract anomalies are a morphological diverse group of developmental disorders. The surgical approach for correction is specific to the type of malformation. Due to the frequent association between müllerian and urinary anomalies, the finding of any of the types should lead to the investigation of the other [2].

Key Words: *Congenital Abnormalities; female genital tract*

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INTRODUCTION

Developmental anomalies of female genital tract are not very common, incidence reported in the literature is 0.2%–3.8% [1]. Congenital uterine malformations result from abnormal formation, fusion, or resorption of the mullerian ducts during fetal life. During their development, they are closely associated with the urinary system. Lateral fusion defects may result in uterus didelphys, bicornuate uterus, or arcuate uterus [3]. Vertical fusion defects results in imperforate hymen, transverse vaginal septum, or oblique vaginal septum. Resorption failure results in a uterus with a partial or complete septum [3]. These abnormalities are often recognized after the onset of puberty. In the pre pubertal period, normal external genitalia and well developed secondary sexual characteristics often mask abnormalities of the internal reproductive organs. The clinical presentation varies from symptoms of primary amenorrhea, obstruction of the menstrual flow in adolescence to hypomenorrhea and fertility problems in adult life. Women with primary amenorrhea, incapacity for vaginal coitus or pelvic pain, due to obstruction of the menstrual flow, are diagnosed earlier because they seek assistance earlier. Women who experience problems related to reproduction, such as recurrent pregnancy loss, premature birth and infertility, are diagnosed during the investigation of these symptoms [4]. The causes of uterine malformations are multifactorial which includes ionizing radiations, viral infections, drug induced and genetic mutations [1]. A thorough knowledge of the development of female genital tract and its associated anomalies is necessary to diagnose these disorders and an adequate workup is imperative before planning the treatment [4].

AIMS AND OBJECTIVES:

The aim of our study is to evaluate the various clinical presentations of congenital female genital tract anomalies, and appropriate management used for the respective cases to improve both obstetric and gynaecological outcomes.

MATERIALS AND METHODS:

A prospective observational study was conducted at the Maharajah's Institute of Medical sciences, Obstetrics and Gynecology department for a period of 3 years (April 2020-March 2023). Among more than 3000 patients reported with

the OPD within the above mentioned time period, 13 cases of genital tract anomalies were diagnosed through clinical history, physical examination, hormonal study, imaging studies and hysterolaparoscopy. The optimum mode of management for each case was individualised and assessed in terms of the final outcome and is being followed-up.

CASE DESCRIPTIONS:

CASE-1:

A 16-year-old girl presented with primary amenorrhea and underdeveloped secondary sexual characters.Her body weight was 44 Kg, height was 154 cm and she hadnormal blood pressure. A physicalexamination revealed no abnormalities. Pubic, axillary hair growth and breast development were scored Tanner stage II. The patient had no evidence of facial dysmorphism, webbing of the neck, or skeletal abnormalities. A 1 cm vagina ending in a blind pouch was found on gynecological examination. Recto vaginal examination revealed no evidence of a cervix or uterus.On ultrasound and MRI, internal genitalia could not be identified and both the ovaries were absent (figure: 1). No associated renal and skeletal anomalies. An endocrine study including pituitary, ovarian, and thyroid evaluation was performed and reveled hypergonadotrophic hypogonadism. Her karyotype was normal, 46 XX. Diagnosed as ovarian agenesis associated with Mullerian agenesis. She was started on oral conjugated estrogens, as hormone substitution therapy remains the only therapeutic option for the development of secondary sexual characteristics and the prevention of osteoporosis. She was counselled regarding future vaginoplasty for sexual function and addressed future fertility options for having children as these two conditions compromise the fertility.

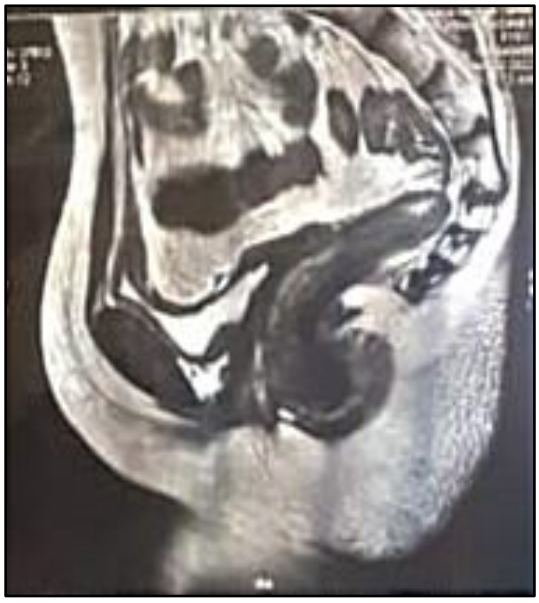

Fig: 1 MRI showing absent internal genitalia and vagina

CASE: 2

A 16-year-old girl presented with primary amenorrhoea but there was no history of cyclic pain abdomen or abdominal lump. Her body weight was 54 Kg, height was 152 cm, and she hadnormal blood pressure. On examination, her secondary sexual characteristics were well developed, there was no abdominal or inguinal swelling, and external genital organs were well developed and the vagina was blind-ending.On an ultrasound and MRI, uterus, cervix, and upper vagina were absent along with agenesis of the left kidney, but bilateral ovaries were normal. Karyotype was 46 XX with a normal hormonal study. Diagnosed as Mullerian agenesis with unilateral renal agenesis (MRKH II). She was counselled regarding vaginoplasty in future and fertility options.

CASE: 3

A 15-year-old girl presented with primary amenorrhea but there was no history of cyclic pain abdomen or abdominal lump. Her body weight was 56 kg, height was 155 cms, and blood pressure was normal. On examination, her secondary sexual characteristics were well developed, there was no abdominal or inguinal swelling, and external genitalia were well developed and the vagina was blind-ending.On an ultrasound and MRI, uterus, cervix, and upper vagina were absent, bilateral ovaries were normal and bilateral kidneys were normal (figure: 2). Karyotype was 46 XX with a normal hormonal study. Since the girl was diagnosed with having Mullerian agenesis (MRKH-1), we counselled her regarding vaginoplasty in future and fertility options.

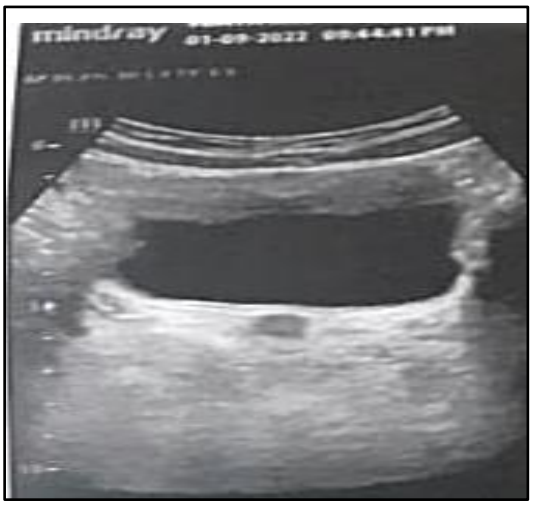

Fig 2: USG shows absent uterus, cervix, and upper vagina

CASE: 4

A 21 years old G2P1D1 with 10 weeks gestation came with complaint of painless bleeding per vaginum. In her 1st pregnancy she delivered an IUGR baby of weight 1.5 kg at 36 weeks of gestation and that babydied within in 24 hours. On examination abdomen was soft and nontender. On speculum examination a vertical vaginal septum and two cervical OS was seen. Some products of conception were seen through left cervical OS. Ultrasound revealed Bicornuate Bicollis uterus with vaginal septum and left uterine horn with gestational sac with no cardiac activity (figure: 3). the left uterine horn is evacuated under anaesthesia.

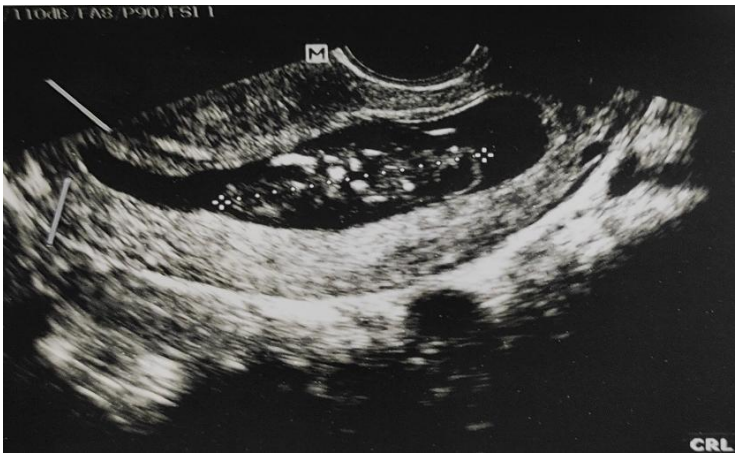

Fig 3: USGshows Bicornuate Bicollis uterus left uterine horn with gestational sac but no cardiac activity

CASE: 5

A 22 year old married nulliparous woman presented with history of inability to conceive. She had been married for two years, and hada complaint of difficulty having sexual intercourse since then. There was no history of cyclical pain in the abdomen or abdominal lump. She attained menarche at 13 years and menstrual cycles were regular 5-6 /30 days, no complaint of dysmenorrhoea. On examination, her secondary sexual characteristics were well developed, there was no abdominal or inguinal swelling, and external genital organs were well developed. On speculum examination a blind vagina is seen with a pin hole opening in the centre (figure: 4). Per rectal examination, uterus was felt and found to be normal in size. An MRI revealed a thin transverse vaginal septum measuring a thickness of 7.5 mm which is 2.1 cm from the introitus with a small fenestration. The proximal and mid portion of the vagina appeared normal. The diagnosis was Transverse vaginal septum, and she underwent resection of vaginal septum under general anaesthesia.

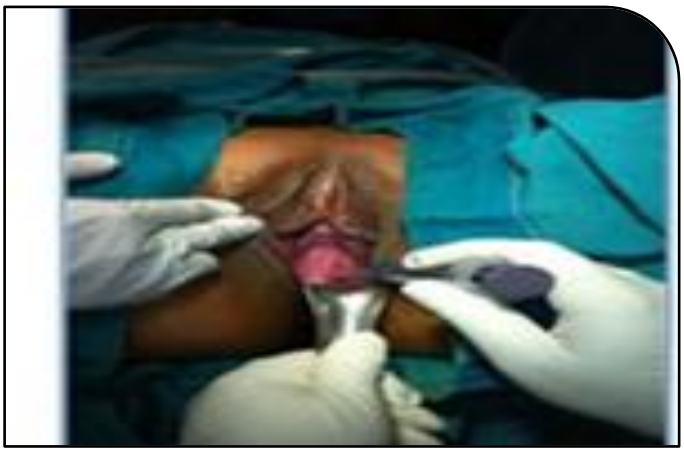

Fig 4: shows a blind vagina with a pin hole opening in the centre

CASE: 6

A 23-year-old nulliparous woman was evaluated for secondary infertility. She had been married for 5 years, and she had three prior spontaneous first trimester miscarriages. All are spontaneous conceptions and confirmed by UPT. Her medical history and menstrual history were not significant. Physical examination revealed well developed secondary sexual characters, normal external genitalia, and no abnormality noted on bi-manual examination. She had been evaluated for recurrent pregnancy loss and report shows a normal ovarian reserve, negative for antiphospholipid antibodies, and hormonal study was normal. The karyotype was 46 XX , and a normal study of husband semen analysis. An HSG, USG & MRI revealed Septate uterus. Hysteroscopic septal resection was done. We reported a successful pregnancy outcome after 2 years.

CASE: 7

A 25 year old G3P1L1A1 at 33 weeks of gestation came with complaint of leaking per vagina. Her obstetric history showed that she had second trimester miscarriage in her first pregnancy. She had a preterm delivery in her second pregnancy at 32 weeks gestation 2 yrs back. She underwent an emergency cesarean section due to fetal distress. Intraoperative findings showed dimple at the uterine fundus, Uterine septum extended from the fundus to the corpus uteri (figure: 5).The Intraoperative and post-operative periods were uneventful.

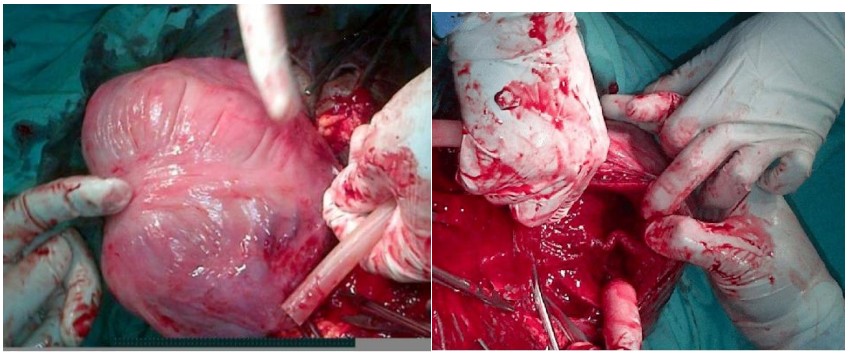

Fig 5: Intra-operative finding: Uterine septum extended from the fundus to the corpus uteri

CASE: 8

A 30 year G2P1L1 was scheduled for an elective cesarean section at 38weeks of gestation. She had cesarean delivery in previous pregnancy due to transverse lie at 39 weeks of pregnancy.USG showed a live intrauterine fetus in transverse lie, placenta was posterior. Intra operative finding, the fetus was in transverse lie and delivery was conducted by internal podalic version and breech extraction. A live male baby that weighed 3.8 Kg was delivered. On examination after the delivery of the baby revealed a uterus with an indentation at the fundus, a diagnosis of Arcuate Uterus was made. The uterine cavity shape and reduction in capacity seen in arcuate uterus could be the reason for the recurrent transverse lie.

CASE: 9

A 22 year primigravida at 37 weeks of gestation presented with pre labour rupture of membranes. Ultrasonography revealed a single live intrauterine gestation of 35 weeks 6 days with breech presentation. In view of the breech presentation, an emergency lower segment cesarean section was done with the following intra operative findings: A live male babyin breech presentation weighing 3.1 kg was delivered. The placenta is located anterior in the upper segment. By exteriorizing the uterus, confirmed the Arcuate Uterus (figure: 6). She had an uneventful post operative period.

Fig 6: Intra-operative finding of Arcuate uterus

CASE: 10

A 42 year old P2L2, with term vaginal deliveries posted for total laparoscopic hysterectomy in view of abnormal uterine bleeding of endometrial cause. No adverse events during pregnancy and delivery were noted. Intraoperatively diagnosed as Arcuate uterus (figure: 7). She had an uneventful post operative period and was discharged on 5th post operative day.

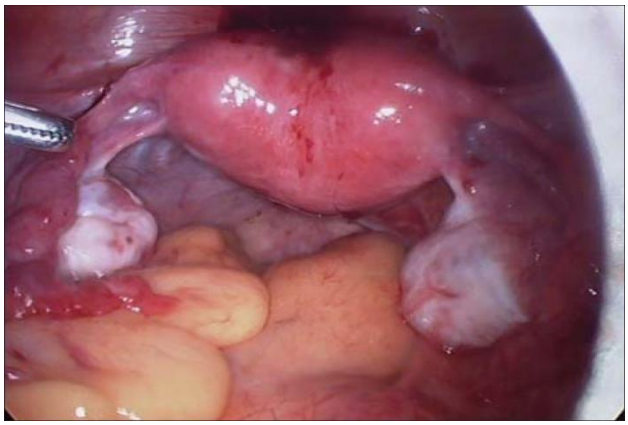

Fig 7: Inoperative finding of laparoscopic hysterectomy: Arcuate uterus

CASE: 11

A 40 year old nulliparous woman came with complaint of lower abdominal pain with stable vitals. She attained menarche at 15 years age and her cycles were regular. An abdominal examination showed themass corresponds to 20 weeks' size of the uterus. Tenderness present on supra pubic, umbilical, right lumbar and iliac regions. On speculum examination blood stained vaginal discharge was seen. Bimanual examination revealed the cervix pointing upwards and deviated to the left. Mass movement is transmitted to the cervix. Bilateral forniceal tenderness was noted.USG showed bulky uterus with multiple fibroids and bilateral enlarged ovaries with endometriotic cyst /tubo-ovarian abscess. Intraoperative findings showed bicornuate uterus with multiple fibroids. Total abdominal hysterectomy with bilateral salpingo oophrectomy with appendicectomy was done.

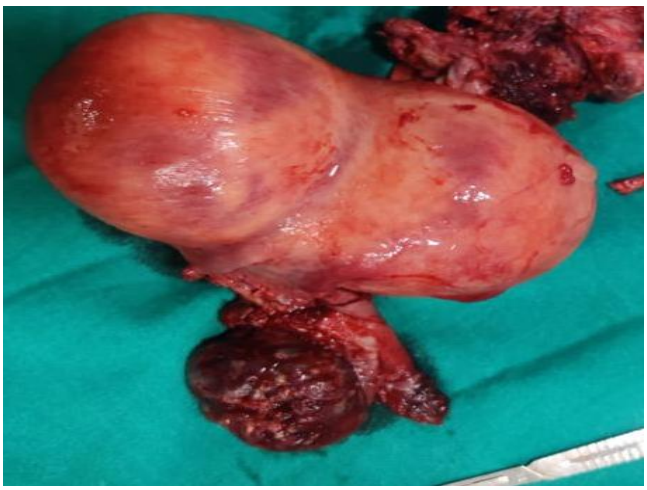

Fig 8: Hysterectomy specimen shows bicornuate uterus with multiple fibroids

CASE: 12

A 16-year-old girl presented with primary amenorrhea and had cyclical abdominal pain for almost a year. At examination we observed a painful mass in the lower abdomen and normal secondary sexual characteristics. Perineal examination showed a bluish bulging hymen. Transabdominal USG, revealed a hypoechoic pelvic mass 12x8 cm behind the bladder, which was suggestive of hematocolpos and hematometra with no other urogenital abnormalities. Diagnosed as imperforate hymen with haematocolpos and haematometra. Under general anesthesia, the patient underwent hymenectomy with a cruciate incision on the obstructing membrane. Approximately 500 ml of dark red blood was drained immediately after the incision. After adequate drainage and irrigation, no other abnormalities were found on examination of the vaginal canal. The abdominal pain resolved completely after the surgery, and she had regular menstruation subsequently. The uterus and vagina were normal in appearance on follow-up sonography.

CASE: 13

14 year old girl presented with complaint of lower abdominal painassociated with tenesmus. There was no abdominal distension but she had observed some suprapubic fullness. She did not have constipation, diarrhoea, vomiting or fever. Shehas not attained menstrual periods but she had developed secondary sexual characteristics. On examination, she was in severe pain, had tender supra pubic mass corresponding to a uterus at 14 weeks. Rectal examination revealed an anterior mass. Perineal examination revealed a bulging imperforate hymen. An abdominal ultrasound done revealed a distended uterus and vagina filled up with homogeneous thick fluid. A diagnosis of hematocolpometra was made. Anx-shaped incision of the hymen was made under anaesthesia and approximately 600ml of thick chocolate coloured blood was evacuated. The edges of the hymen were everted and anchored by vicryl 2/0 sutures. She made an uneventful recovery, and, however, lost follow-up after that.

Table 1: The Clinical Picture and Associated Anomaly

S. NO.	CLINICAL PRESENTATION	DIAGNOSIS
Case-1	A 15 year old girl with primary amennorhea with under developed secondary sexual characters	Ovarian agenesis associated with Mullerian agenesis.
Case-2	A 16 year old with primary amennorhea with well developed secondary sexual characters	Mullerian agenesis with unilateral renal agenesis(MRKH II)
Case-3	A 15 yearold with well developed secondary sexual characteristics	Mullerian agenesis (MRKH-1)
Case-4	A 21 year G2P1D1 old with 10 weeks gestation with bleeding per vagina.	Bicornuate Bicollis uterus with vaginal septum
Case-5	A 22 year old with primary infertility and dyspareunia	Transverse vaginal septum
Case-6	A 23 years old nulliparous woman with secondary infertility	Septate uterus
Case-7	A 25 year old G3P1L1A1 with h/o preterm delivery and second trimester abortion	Septate uterus
Case-8	A 33 year old G2P1L1 with 38weeks GA with PCP in transverse lie	Arcuate uterus
Case-9	A 22year old primi with 37 weeks GA with PROM in breech presentation	Arcuate uterus
Case-10	A 42 year old P2L2withAUB-E posted for hysterectomy	Arcuate uterus
Case-11	A 40 year old nulliparous woman with complaint of lower abdominal pain with mass per abdomen	Bicornuate uterus with multiple fibroids
Case-12	A 16-year-old girl with primary amenorrhea and had cyclical abdominal pain for a year	Imperforate hymen
Case-13	A 15-year-old girl with primary amenorrhea and acute pain abdomen.	Imperforate hymen

DISCUSSION

In the present study out of approximately 3000 cases, 13 cases were found to be with mullerian abnormalities. Commonest presenting complaint was primary amenorrhea. Most of them presented at an early age group of 15– 25 yrs. Theincidence of mullerian anomalies reported in the literature is 0.2%–3.8% [1]. According to Golan A et al, Strassmann EO et al, incidence of mullerian anomalies is 0.1%–3.5% [5, 6]. Present study shows prevalence of mullerian anomalies is 0.4% at our tertiary care center. Kabadi, Y.M et al [1] has been reported that rates of cesarean delivery are increased in cases of uterine anomalies, this finding is similar to our study. Akhtar MA et al reported, septate uterus is the most common mullerian anomaly, and is associated with high rates of both first- and second-trimester miscarriage, preterm delivery, and fetal malpresentation [7], this finding is similar to our study.

CONCLUSION

Mullerian anomalies are a morphological diverse group of developmental disorders. A careful history and thorough clinical examination plays a fundamental role. Due to the frequent association between müllerian and urinary anomalies, the finding of any of the types should lead to the investigation of the other. Management must also be individualised considering anatomical and clinical characteristics and the patient's wishes.

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REFERENCES

1. Kabadi, Y.M., Ayyanar, A. (2022). A "cluster" of ten uterine anomalies observed in a single center over a short period of 4 weeks: a case series. *J Med Case Reports*. 16, 130
2. Nishu, D.S., Uddin, M.M., Akter, K. et al. (2019). Herlyn-Werner-Wunderlich syndrome presenting with dysmenorrhea: a case report. *J Med Case Reports*. **13**, 323. https://doi.org/10.1186/s13256-019-2258-6
3. Kansara M, Gothwal M, Yadav G, Singh P, Kathuriya P. (2022). Mullerian anomalies showing a variety of clinical scenarios: case series. *Glob J Gynecol Obstet Res*. 1(1):1–6
4. Gupta U, Tripathi V, Sharma P. (2021). Clinical Implications and ESHRE/ESGE Classification of Mullerian Anomalies: A Case Series. *Obstet Gynecol Cases Rev*. 8:191.
5. Golan A, Langer R, Bukovsky I, Caspi E. (1989). Congenital anomalies of the müllerian system. Fertil Steril. 51(5):747–755.
6. Strassmann EO. (1966). Fertility and unification of double uterus. Fertil Steril. 17(2):165–176.
7. Akhtar MA, Saravelos SH, Li TC, Jayaprakasan K; (2020). Royal College of Obstetricians and Gynaecologists. Reproductive Implications and Management of Congenital Uterine Anomalies: Scientific Impact Paper No.62 November 2019. BJOG. 127(5):e1–e13.