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Case Snippets

Dr. Aswathkumar
President KFOG

Dr. Venugopal
Secretary KFOG

PRESIDENT'S MESSAGE

Seasons greetings to all KFOGians!

My sincere gratitude to all of you for giving me the opportunity to serve as the President of the Kerala federation of Obstetrics and Gynecology. The turbulent COVID times has shown us the need for continued medical education. It gives me immense pleasure to introduce the inaugural issue of KFOG Case Snippets which shall showcase relevant cases of practical importance.

I congratulate Dr. Shyama Devadasan, Editor for meticulously compiling the challenging cases and bringing out an exemplary edition to update the knowledge of our members.

Warm regards

Dr. Aswath Kumar
President KFOG (2023-24)

EDITOR'S MESSAGE

"Wisdom.... comes not from age, but from education and learning."
- Anton Chekhov

It is a great honour to assume the role of Editor of this new endeavour under the able leadership of our President, Dr. Aswath Kumar. The KFOG Case Snippets will be released bimonthly to exclusively publish case reports on diverse topics in OBG. I thank each one of the authors, for their timely submissions to the journal. I warmly welcome the participation and support of every reader with their valuable contributions. Kindly send them along with any queries to drshyamadevadasan12@gmail.com.

With best wishes for the year ahead,

Dr. Shyama Devadasan
Editor, KFOG Case Snippets

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A UNIQUE CASE OF RECURRENT GESTATIONAL TROPHOBLASTIC DISEASE WITH HOMOZYGOUS PATHOGENIC MUTATION IN NLRP7 GENE

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INTRODUCTION

Gestational trophoblastic disease (GTD) is a spectrum of conditions that arises due to abnormal proliferation of trophoblastic tissue which is foreign to mother. It includes:

- **Benign-**
  Vesicular(hydatidiform) mole
  Placental site nodule
  Exaggerated placental site
- **Malignant-**
  Invasive mole
  Choriocarcinoma
  Placental site trophoblastic tumour
  Epithelioid trophoblastic tumour

There are two types of hydatidiform mole namely complete and partial.

Complete hydatidiform mole results from the fertilization of an egg, from which the nuclear material has been lost or inactivated, by a single sperm being 23x chromosomes which duplicates to 46xx. This makes complete mole homozygous. Less frequently, fertilization occurs with two sperms resulting in either 46xx or 46xy heterozygous chromosomal constitution.
In partial hydatidiform mole, maternal chromosomes are present and the condition arises by diandry (one maternal and two paternal sets of chromosomes).
The risk of recurrence of hydatidiform mole is 0.5-2.8% with a subsequent greater risk of developing invasive mole or choriocarcinoma. The risk of repeat hydatidiform mole in next pregnancy is 1:76 while the risk after two past hydatidiform mole is 1:6.5 and after three previous molar pregnancies it is almost 100%. Two maternal-effect genes, NLRP7 and KHDC3L are identified to have causal roles in recurrent hydatidiform mole.
A case of molar pregnancy, recurring for the third consecutive time is reported.

CASE PRESENTATION

A 28 year old , G4A3, with 7 weeks of amenorrhea and positive urine pregnancy test came with complaints of bleeding per vaginum.

Obstetric history

A1-?Biochemical Pregnancy

A2- USG-?molar pregnancy
Suction and evacuation done at 8 weeks and diagnosis confirmed by histopathological examination.
β HCG regressed spontaneously.

A3- USG-?molar pregnancy
Suction and evacuation done at 10 weeks and diagnosis confirmed by histopathological examination.

β HCG regressed after methotrexate injection.
G4-Present pregnancy
There is no relevant past or family history.

Clinical examination:
She moderately built and nourished. Her vitals were stable. On examination, her abdomen was soft, non tender with no palpable mass. Per speculum examination revealed a healthy cervix, bleeding was noted. Bimanual pelvic examination showed a mobile uterus of 8-10 weeks size, fornices free.

Investigations-
- Transvaginal sonography: Uterus 8.2x4.5x3.2cm, Endometrium-showing cystic spaces in the uterine cavity.Right ovary-corpus luteal cyst of size 20mm. Left ovary normal. No adnexal masses.

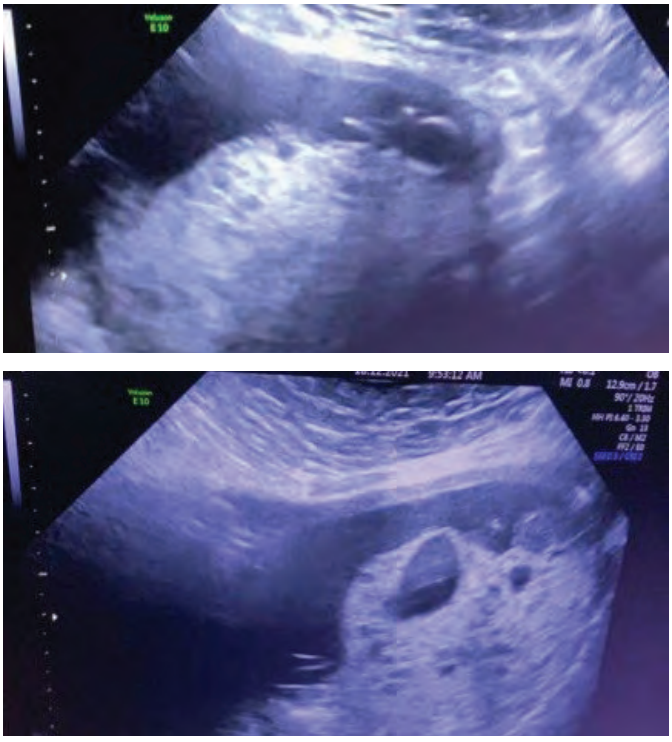

Figure 1 and 2. Ultrasound images showing cystic changes in the uterine endometrium

- Serum B-HCG - 22587IU/ml.

A Molar pregnancy was diagnosed and evacuated under anesthesia. Products of conception were sent for histopathological examination. After 1 week, the β HCG was reported as 47642 IU/ml. An USG revealed retained products of conception of 35mm. She was taken for second round of evacuation.

The products of conception was sent for histopathological examination and a genetic workup. She was given 8 cycles of Inj Methotrexate post evacuation and β HCG regressed.

Histopathology results:
- Degenerating products of conception with focal hydropic changes.
- Karyotype: 46XY

Genetic workup results:
- Targeted gene sequencing showed homozygous pathogenic mutation of NLRP7(NOD Like Receptor Pyrin containing 7) gene
- Single base pair deletion in Exon 6 of the NLRP7 gene- c.1603delG(p.Glu535ArgfsT er3)

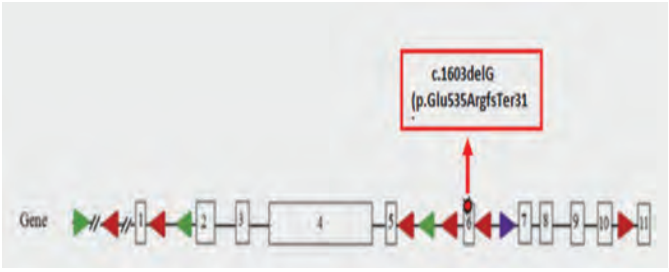

Figure 3. Showing NLRP7 gene structure. Arrow indicates mutation (deletion) in Exon 6 at position c.1603delG(p.Glu535ArgfsT er3)

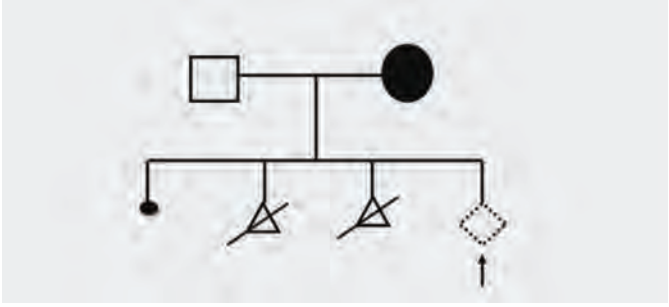

Figure 4. Shaded symbol shows affected individual

In this regard , the couple was counselled regarding IVF with donor oocyte in order to obtain a normal pregnancy. She conceived after IVF – OD, with DCDA twins which was delivered at 36 weeks.
The examination of placenta, membranes and umbilical cord revealed normal histology.

DISCUSSION

Among the GTDs, the most common is Hydatidiform Mole (HM), and it is the only one that has been reported to recur, which indicates the patients' genetic predisposition. In addition, HM is the only GTD that segregates in families according to

Mendel's laws of heredity, which made it possible to use rare familial cases of recurrent HMs (RHMs) to identify two maternal-effect genes, NLRP7 and KHDC3L, responsible for this condition.

NLRP7, a nucleotide oligomerization domain (NOD)-like receptor, pyrin containing 7, maps to 19q13.4 and is the first identified causative gene for RHMs. It's overexpression in transient transfections downregulates the production of IL-1β, an important mediator of the inflammatory response. Another role for NLRP7 which was recently demonstrated was that NLRP7 knockdown in human embryonic stem cells led to an earlier expression of two trophoblast differentiation markers, GCM1 and INSL4, suggesting that NLRP7 loss of function accelerates trophoblast differentiation.

NLRP7 knockdown also increased the level of human chorionic gonadotropin (hCG), known to be very high in patients with molar pregnancies. Individually, none of the above-described roles of NLRP7 may explain entirely the pathology of moles. Perhaps a combination of the above-described functions, with some acting in the oocytes and affecting the differentiation and proliferation of embryonic and trophoblastic tissues, and others acting in hematopoietic inflammatory cells present in the endometrium and downregulating the maternal immune response, together contribute to the three fundamental aspects of moles: retained human pregnancies with no embryo and excessive trophoblastic proliferation.

KHDC3L (KH domain containing 3-like), which was identified in 2011, is a second recessive gene responsible for RHMs. KHDC3L maps to chromosome 6, and available data indicate that this gene is a minor gene for RHMs, accounting for 10–14 % of patients who do not have mutations in NLRP7.

Because of the high rate of NLRP7 mutations in patients with RHMs, which seems to vary with populations, patients with at least two HMs (complete or partial) should be first offered NLRP7 DNA testing. Patients without NLRP7 mutations should be screened for KHDC3L mutations.

Prognosis for Future Pregnancies

The goal of patients seeking DNA testing is to ascertain their chances of conceiving healthy babies and their risk for mole recurrence and malignant sequelae. Studies from various groups have shown that the chances of a normal live birth are very low in women with mutations in NLRP7 or KHDC3L. Based on available data, both genes are required in the oocytes. Therefore, oocyte donation is the option for further pregnancy.

CONCLUSION

This reveals the importance of genetic workup among patients with recurrent hydatidiform mole to know the association between recurrent gestational trophoblastic disease and pathogenic mutation. This helps in proper planning of future conception.

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02

DOUBLE TROUBLE- A CASE OF SPONTANEOUS HETEROTOPIC PREGNANCY

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INTRODUCTION

Heterotopic pregnancy (HP) was first described in 1708 by Duverney. It is defined as a rare condition when intrauterine and extrauterine gestations coexist. The theoretically calculated incidence of a spontaneous HP is approximately 1 in 30,000 [1], while some risk factors increase the frequency up to 1 in 100 [2,3,4]. HP is a life-threatening condition that may lead to complications such as the rupture of an ectopic pregnancy (EP) and the loss of the intrauterine (IU) embryo after treatment [5].

We report a case of a low risk pregnancy ,spontaneous conception ,heterotopic pregnancy in a 28-year-old patient with the successful treatment of an ectopic pregnancy with the preservation of fallopian tubes and the IU embryo, which resulted in a spontaneous delivery.

CASE PRESENTATION

A 28 year old otherwise healthy women (G3P2L2) at her seventh week of gestation presented to the emergency department complaining of weakness and acute pain in lower abdomen and epigastric region. The pain had lasted for one day without nausea, vomiting, or other gastrointestinal symptoms. The patient did not have any vaginal bleeding and denied any other illnesses or allergies. A physical examination revealed a normal body temperature and arterial blood pressure (110/60 mmHg), along with tachycardia (110 beats per minute) and pain of the lower abdomen during palpation. Laboratory testing on admission showed an elevated white blood cell count up to 16000,serum hemoglobin concentration of 8.5g along with a normal blood platelet level of 329 × 109/L. The serum β-human chorionic gonadotropin (β-hCG) level was 75,635 U/L. Transvaginal ultrasonography (TVUS) revealed intrauterine (IU) gestation with a sac of 25.2 mm in diameter and a crown–rump length (CRL) of 11.2 mm with a positive embryo heart rate and an extrauterine gestation in the right fallopian tube with a sac of 20.2 mm and a CRL of 13.7 mm

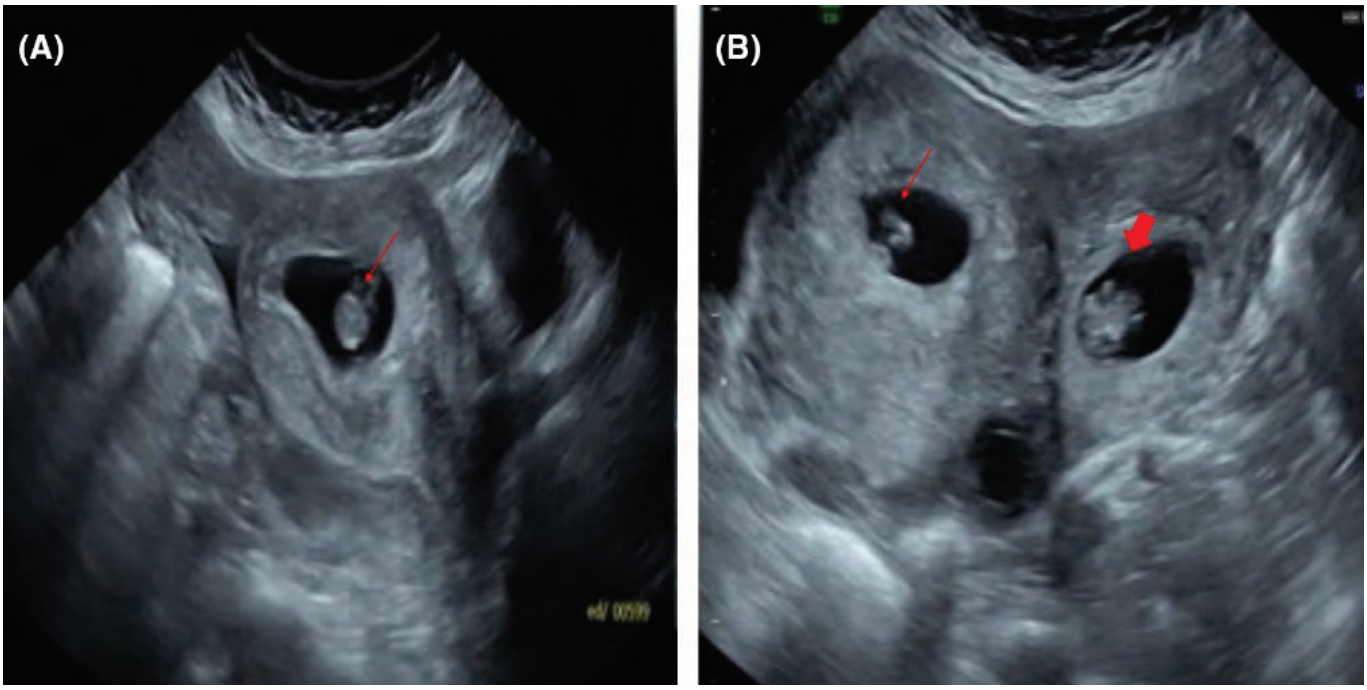

Figure A & B. Transvaginal ultrasonography (TVUS) image of the uterus (transverse view) showing an intrauterine gestation (left) coexisting with an ectopic tubal pregnancy (left) with a sac containing an embryo with a crown rump length of 13.7 mm.

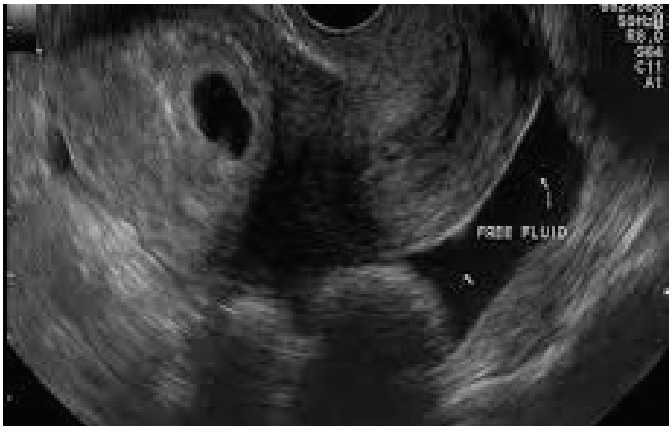

Figure C : TVUS image showing free intraperitoneal fluid in pelvis.

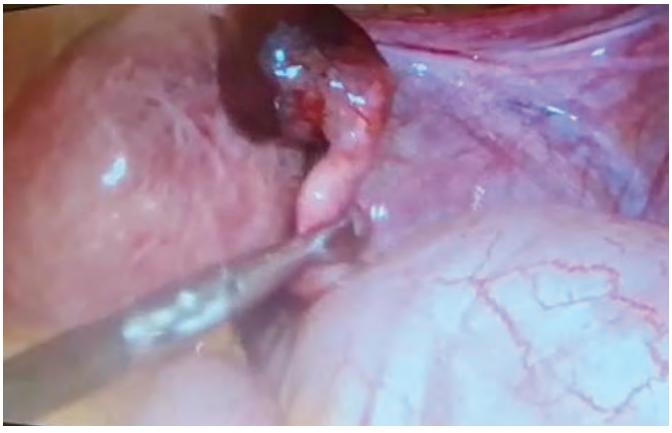

Figure D: Ectopic mass noted in laparoscopy

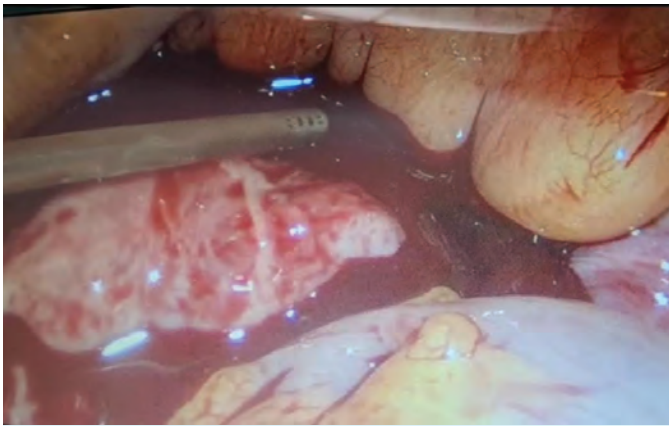

Figure E: Hemoperitoneum noted in laparoscopy

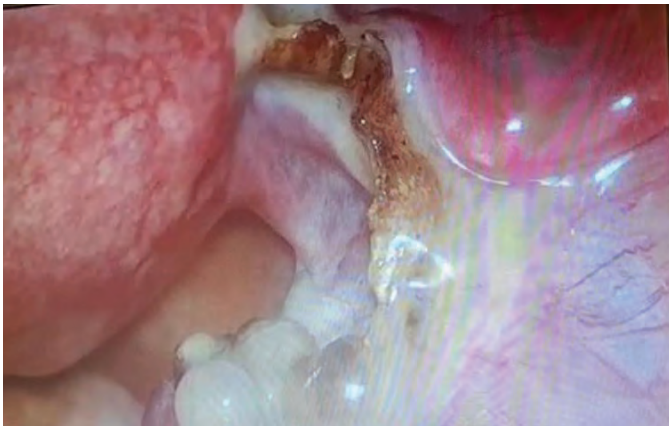

Figure F: Post surgical view in laparoscopy

with cardiac activity (Figure A & B). TVUS also demonstrated free intraperitoneal fluid in the pouch of douglas (Figure C). An urgent right laparoscopic salpingotomy was performed under general anesthesia, 0.5 L of blood was evacuated from the free peritoneal cavity, and the ectopic embryo was found in the right fallopian tube (Figure D and E) . The ectopic sac was successfully removed. A histological examination confirmed chorionic villi suggestive of an ectopic pregnancy.

There were no complications during the post operative course. After the surgery, she was put on progesterone support (200 mg/day intravaginally) and was continued until 12-weeks of gestation. The patient recovered well and was discharged from the hospital on the third postoperative day. She had regular antenatal care appointments, and her pregnancy was uncomplicated. The development of the fetus was normal and eventually, at 39 weeks and four days of gestation, the patient gave vaginal birth to a healthy male baby 54 cm tall and weighed 3280 g. Postnatal recovery was without any complications, and the patient was discharged on the third postpartum day.

3. DISCUSSION

Heterotopic pregnancy is defined as a multiple gestation with one embryo inside the uterus and the other one elsewhere. This condition has become more common and relevant because of widespread assisted reproductive technologies (ARTs) and ovarian stimulation for infertility treatment [2,3]. Other risk factors for HP are pelvic inflammatory disease (PID), pelvic surgery, and previous fallopian tube damage or pathology [3]. Our patient did not have any of these risk factors and conceived spontaneously, which makes this case very rare and hard to detect.

In 95% of cases, the EP occurs in the fallopian tube [6], but it can also be found in the cervix, scar from a prior cesarean section, and the interstitial segment of a fallopian tube, ovary, peritoneal, or abdominal cavity [7]. The apparent increase in the incidence of nontubal EPs including HP may be attributed to the higher number of pregnancies after in vitro fertilization treatment [6]. Our case described a case of a spontaneous conception,uncomplicated pregnancy with heterotopic pregnancy.

Tal J et al. reported that 70% of all HP cases are diagnosed between five and eight weeks of gestation, 20% between 9 and 10 weeks, and only 10% after the 11th week [2]. The symptoms of HP are nonspecific. HP can be asymptomatic in 24% of cases [4,8]. Abdominal pain is the most frequent symptom of HP, though vaginal bleeding and hypovolemic shock are also common [4,8]. Vaginal bleeding and hypovolemic shock often indicate the rupture of the EP and require urgent treatment. Our patient was admitted to the emergency room complaining of the pain in the abdomen with no other symptoms, which made a differential diagnosis difficult.

The early diagnosis of HP is challenging because a raised serum β-hCG level and an intrauterine embryo seen on US lead one to think about normal pregnancy, and almost no one examines for an EP if the patient is asymptomatic. When an intrauterine embryo is found, it is crucial to inspect the adnexa of the uterus and to record it. The identification of an EP on USG has a reported sensitivity and specificity of 71–100% and 41–99%, respectively [9]. Combined serum β-hCG measurement and TVUS improve the diagnostic sensitivity of HP [3]. TVUS has been found to be better in early diagnosis compared to transabdominal US. It detects almost 70% of cases between the fifth and eighth weeks of gestation [10]. In this case, both a serum β-hCG measurement and TVUS were done at the emergency room, and HP was suspected because both embryos were visualized and one of them was outside the uterus.

Treatment possibilities include expectant management, surgical management (either laparoscopy or laparotomy). Treatment depends on the patient's condition, the size and site of an EP, previous pregnancies, the viability of intrauterine and extrauterine gestation, and the expertise of the physicians. Expectant management can be selected in symptom-free patients where the unruptured ectopic embryo has a limited craniocaudal length with no cardiac activity [11].

In our case, an urgent right laparoscopic salpingotomy was chosen due to the free intraperitoneal fluid in the lesser pelvis and the suspicion of the rupture of the EP. The postoperative period was successful with normal growth of the IU embryo. The left fallopian tube was preserved, which is extremely important for the young woman for her future fertility About 60–70% of HP cases result in live childbirth with outcomes similar to that of singleton pregnancies [4].

4. CONCLUSIONS

All pregnant women presenting with abdominal pain or vaginal bleeding should be suspected of heterotopic pregnancy even if the conception is spontaneous. Combined serum β-hCG measurements and TVUS are efficient for diagnosing HP. During ultrasound examination, it is very important to check the uterus, adnexa and pouch of douglas. Treatment options include different methods from observation to surgery and should be chosen depending on the clinical situation. If diagnosed and treated on time, heterotopic pregnancy has favourable outcomes for intrauterine pregnancy and the woman. In this case, the need for luteal phase support for a good outcome remains unclear due to a lack of evidence.

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03

A CASE OF SPONTANEOUS UTERINE RUPTURE IN A PRIMIGRAVIDA

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ABSTRACT

Uterine rupture is a rare but disastrous obstetric complication, associated with high rates of maternal and perinatal morbidity and mortality (1). Even though rupture is more common in a scarred uterus, uterine rupture in a primigravida has also been reported. History of any uterine surgeries like previous cesarean section, myomectomy or partial uterine resection is a recognized risk factor for uterine rupture during labor. Uterine rupture is extremely rare prior to the onset of labor and in nulliparous women (2).

Here we present a case report of a spontaneous uterine rupture in a primigravida in preterm labor, with a history of laparoscopic "cystectomy" in the past.

CASE REPORT

A 36-year-old primigravida with a married life of 10 years and a long period of infertility, now with a spontaneous conception is at 33 weeks and 1 day gestation. Her medical history was noteworthy for she had tried a few cycles of ovulation induction and intrauterine insemination, and later was diagnosed with endometriosis. She then underwent in vitro fertilization at the age of 31 years but did not conceive. After 4 years of the failed IVF, she underwent laparoscopic cystectomy (details of which were not available) and she conceived spontaneously 9 months after the surgery. The pregnancy was uneventful till she presented with intermittent abdominal pain at the clinic where she had her regular antenatal check-ups. Cardiotocography showed one deceleration (FHR dropped up to 80 bpm) and she was referred to our tertiary hospital for further management.

On admission to the labor room, the patient was conscious and had abdominal pain. She was perceiving good fetal movements. Clinical evaluation on admission showed an unfavorable cervix and a single viable fetus. Laboratory evaluation revealed normal findings. CTG taken showed a baseline FHR

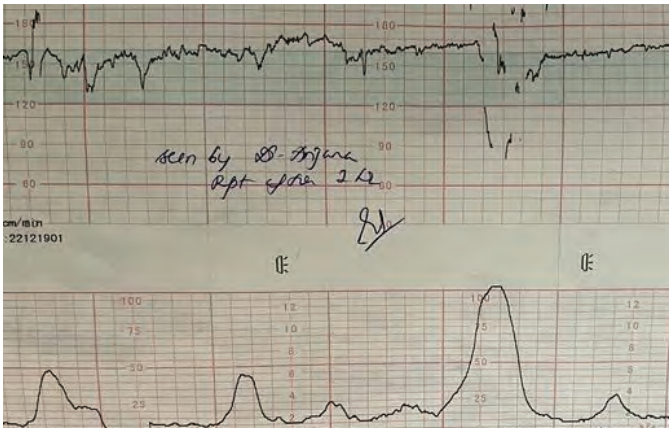

Figure 1: Cardiotocograph on admission

of 160 bpm. She was having uterine contractions (2 contractions lasting for 20-30 seconds in 10 minutes). She was started on tocolytics and posted for antenatal USG for fetal well-being and was given antenatal corticosteroid for fetal lung maturation.

One and half hours after admission the patient suddenly complained of severe abdominal pain. Blood pressure dropped to 90/70 mm Hg with persistent maternal tachycardia. The patient became very pale and drowsy. Fetal doppler showed fetal bradycardia. The patient was immediately shifted to the operation theatre and underwent emergency laparotomy under general anesthesia, for fetal distress and suspected uterine rupture. On opening the abdomen, a massive hemoperitoneum with a large 10 cm fundal rupture was seen. The placenta was seen extruding into the abdominal cavity. A live non-vigorous baby was delivered through the rent itself, with an APGAR score of 3 at 1 minute, and was handed over to the neonatologist. The infant was intubated and connected to mechanical ventilation and shifted to NICU.

After evacuating the hemoperitoneum, the uterus was examined and showed dense posterior wall bowel adhesions and a 10 cm fundal rent. The uterine defect was repaired in three layers with 1-0 synthetic, absorbable suture material.

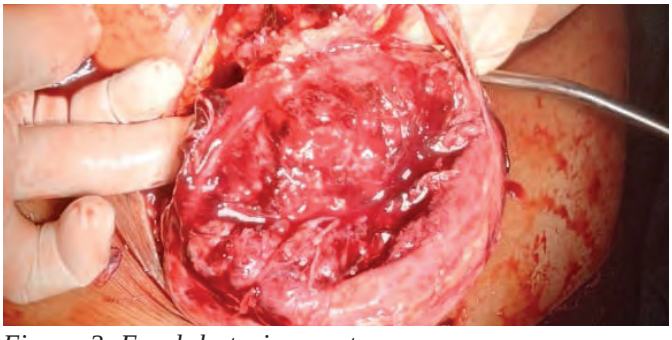

Figure 2: Fundal uterine rupture

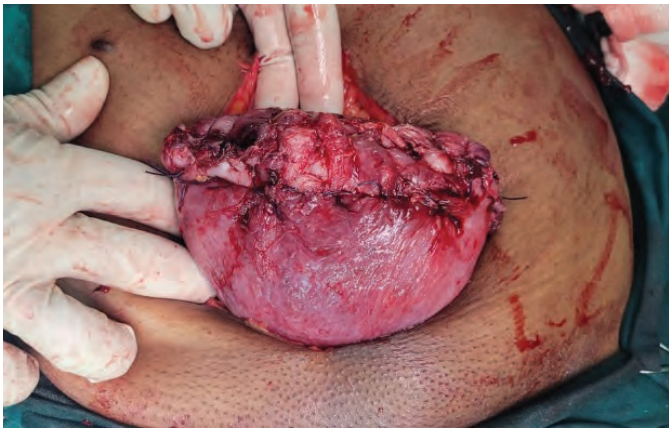

Figure 3: Fundal rupture sutured in layers

Posterior adhesions were released by sharp dissection and small bowel tears were sutured with 3-0 absorbable suture material.

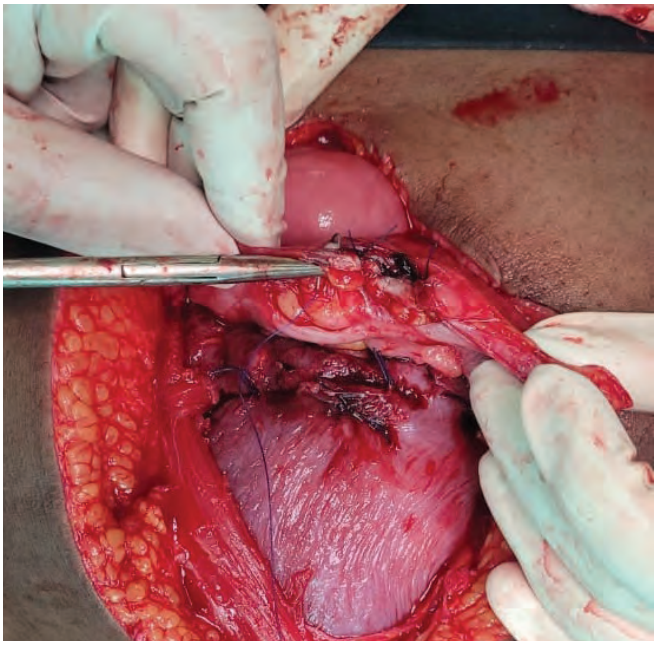

Figure 4: Bowel tears sutured with 3-0 vicryl

Total blood loss was estimated to be 2000 ml and the patient was transfused with 1 unit of packed red cells intraoperatively. The abdomen was closed after keeping an intraperitoneal drain and the patient was shifted to the critical care unit for post-operative monitoring.

The infant required high ventilatory support and developed neonatal seizures in the second hour of life. Later the infant also developed pulmonary artery hypertension, cardiac failure, and prerenal failure.

DISCUSSION

This is a case of rupture in a primigravida with no history of any uterine surgeries in the past. Even though the nulliparous, unscarred uterus is thought to be immune to rupture, (2) in this case, a rupture was suspected as the patient had abdominal pain, tachycardia and hypotension with the presence of fetal bradycardia.

A case reported by Welsh et al (3) was of a spontaneous uterine rupture in a primigravida at 35 weeks who had undergone an "ovarian cystectomy" three years prior. The authors speculated that this rupture was likely secondary to a scarred uterus from an unrevealed termination via hysterotomy, a myomectomy, or possible uterine trauma at the time of the cystectomy. Some documented risk factors of uterine rupture in nulliparous women include previous undisclosed surgical termination of pregnancy, myomectomy, and myolysis (4). Uterine perforation during operative hysteroscopy with the use of excisional electrosurgical procedures has been associated with uterine rupture in subsequent pregnancies (5).

In another review, it was noted that 52.5% of primigravidas with rupture had a history of infertility. The authors concluded that iatrogenic uterine damage could be the cause for uterine rupture in a primigravida, as most infertile patients frequently undergo diagnostic or operative procedures on their uterus during diagnostic investigation or treatment (6).

Maternal and perinatal morbidity and mortality are high in cases of rupture uterus and management of rupture uterus depends on the type, site, and extent of rupture and should be tailored to the patient. Both maternal and fetal mortality rates are more in a case of a rupture in an unscarred uterus when compared to a scarred one (3).

Regarding our case, we were unable to determine a definite cause for rupture, but we hypothesize that iatrogenic uterine injury which might have occurred at the time of laparoscopic cystectomy could be the cause for the rupture when our patient got into preterm labor.

CONCLUSION

Even though uterine rupture in a nullipara with no history of uterine surgeries is a rare complication, especially in this era of increasing myomectomies and infertility treatments, only high clinical suspicion and early diagnosis could aid in improving both maternal and neonatal outcomes.

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04

AN UNUSUAL CASE OF XY GONADAL DYSGENESIS WITHOUT UTERUS

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INTRODUCTION

Mixed gonadal dysgenesis (MGD) is a condition of unusual and asymmetrical gonadal development leading to an unassigned sex differentiation. A number of differences have been reported in the karyotype, most commonly a mosaicism 45,X/ 46, XY. We report an interesting case of mixed gonadal dysgenesis (1,2).

CASE PRESENTATION:

Miss XYZ, a 22-year-old unmarried girl presented with primary amenorrhea to our department in 2021. At 14 years, she had consulted a doctor with primary amenorrhea, minimal development of breasts and absent pubic and axillary hair. She was initially given progesterone tablets but did not have any withdrawal bleeding. From 16 years, she had been taking Ayurvedic treatment on and off. But neither did she attain menarche nor did she have breast, pubic or axillary hair development. Hence at 19 years, she again consulted a gynaecologist when she was further evaluated. Serum hormonal assay and ultrasound were done which showed increased FSH and hypoplastic uterus according to the patient but no documents were available. Then she was given continuous low dose estrogen (Premarin 0.325 mg daily) for 2 years. After 1 year, breast & pubic hair started developing, axillary hair remained scanty, but she did not attain menarche. Hence, she came to our department in 2021.

Past and Family History:

She is the first child of a non-consanguineous marriage. She was born out of a full-term emergency lower segment caesarean section done in view of pre-eclamptic toxemia and had a birth weight of 2.08 kg. She had undergone surgery for bilateral CTEV and bilateral congenital cataracts by 2 and 5 years of age respectively. She had no developmental delays. There was no history of any abdominal, pelvic or genital surgery. She has a younger male sibling, who has normal development for his age. There is no family history of delayed puberty, primary amenorrhea or premature menopause. There is no history of genetic defects, delayed milestones, mental retardation or infertility in the family.

Clinical Examination:

She is moderately built and nourished. She has a height of 164 cm, weight of 70 kg and BMI of 26. Axillary hair was scanty; breast and pubic hair development were of Tanner Stage 4 and 3 respectively. On genital examination: labia majora appeared normal, labia minora were fused with a central opening and clitoris measured 12 mm in length. Urethra and vagina were not seen separately while anal opening was normal. On per vaginal examination, vaginal introitus admits just one finger. On per rectal examination, no uterus was palpable.

Investigations:

Hormonal assay: FSH - 80 microIU/ml; AMH - 0.03 ng/ml; Testosterone - 1.35 ng/dl; DHEAS - 60 micrograms/dl (134-407 micrograms/dl); PRL - 18.4 ng/dl.

Karyotype: 46 XY

MRI Abdomen & Pelvis: No uterus seen; a blind

ending lower vaginal canal measuring 3.54 cm in length seen; small cystic regions seen in bilateral proximal inguinal canals measuring 13.4 mm on right side and 7.9 mm on left side probably free fluid/cysts/streak gonads with cystic degeneration; bilateral kidneys normal in size and morphology.

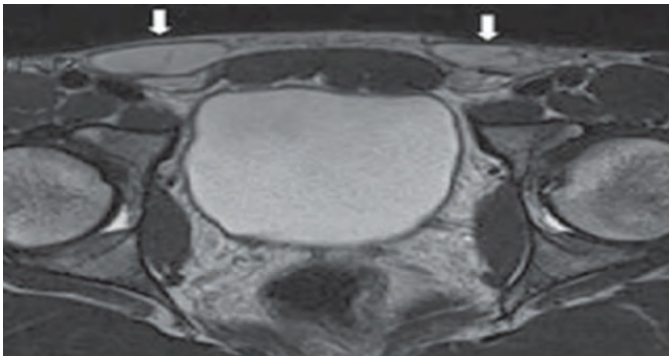

Figure 1. MRI Pelvis showing bilateral streak gonads

Management:

Based on history, clinical examination and investigations done, a provisional diagnosis of partial XY gonadal dysgenesis was made. After counselling about the condition, its implications and the follow up required, it was decided to proceed with bilateral gonadectomy.

Laparoscopy: 3 laparoscopic ports were placed, insufflation done with 2 litres of CO2 and pneumoperitoneum was created. Uterus & tubes were absent and both gonads (testes) appeared atrophic & were seen at the proximal end of the inguinal canal.

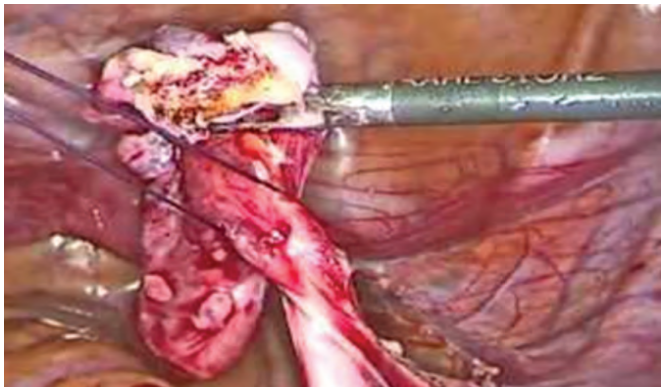

Figure 2. Streak testes on laparoscopy

Hysteroscopy: A small vaginal pouch was seen but urethra was not seen clearly. Hysteroscope was inserted into vaginal pouch and urethra was visualized in the anterior part of the vestibule. Then the hysteroscope was inserted into the urethra also and both ureteric orifices were identified.

Both testes were removed after isolating and cauterising the testicular vessels and thus bilateral orchidectomy was completed and the specimens obtained were sent for histopathological examination.

Histopathology:

Peritoneal fluid cytology: a few lymphocytes in a background of eosinophilic material with no atypical cells; *Macroscopic*: right testis – grey white brownish tissue 4.5 X 1.5 X 1 cm; left testis – grey white brownish tissue 3.1 X 0.5 cm; Microscopic: tubules of varying sizes lined by pseudostratified ciliated columnar epithelium with congested dilated blood vessels and areas of haemorrhage and adipocytes suggestive of epididymal tissue.

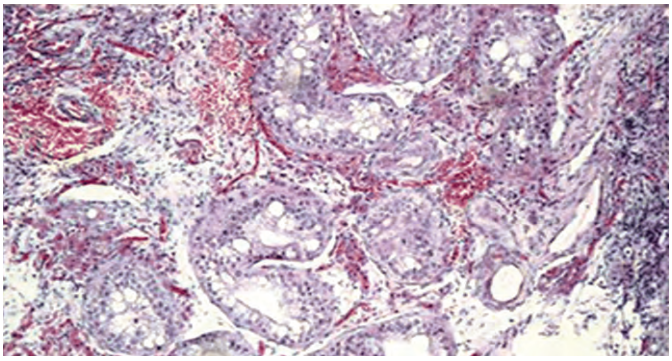

Figure 3. Pseudostratified ciliated columnar epithelium on HPE

A final diagnosis of partial XY gonadal dysgenesis was made after the intraoperative and histopathological findings.

DISCUSSION:

In this case, a diagnosis of partial 46 XY gonadal dysgenesis was made where Mullerian structures may be present or absent and external genitalia may be female, male or ambiguous. Also, there can be wide variations in the phenotype associated with other developmental abnormalities. Complete/ Pure 46 XY gonadal dysgenesis (Swyer's syndrome) is characterised by dysgenetic streak gonads that produce neither anti mullerian hormone nor androgens. Consequently, female internal genitalia develop and internal and external genitalia do not masculinise resulting in a female phenotype with primary amenorrhea. This can also be a variant of Swyer's syndrome without uterus according to literature. Another differential diagnosis is Testicular Regression Syndrome which has a similar histopathological finding but that is characterized by partial or complete absence of testicular tissue in the presence of normal male external genitalia. Laparoscopic bilateral gonadectomy should be done soon after diagnosis as risk of malignant germ cell tumours arising from dysgenetic gonads is around 25%. Post operative hormone replacement therapy with conjugated estrogen is necessary for bone & fe-

male secondary sexual characteristics development. But she can neither conceive due to absence of ovaries nor bear a child as she does not have a uterus (3).

CONCLUSION

MGD represents an intermediate between pure gonadal dysgenesis and ovotesticular DSD (OT-DSD). These patients usually need a multidisciplinary approach for management, but still long-term data on fertility and functional improvement of each approach of treatment is required.

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