Case Report

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Sirenomelia

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Abstract

Sirenomelia (mermaid syndrome) is a rare congenital anomaly with characteristic feature of complete or partial fusion of lower limbs. Although, this syndrome is incompatible with life due to the association of several congenital visceral abnormalities; however, there are few reports of surviving infants [1]. Here, we are reporting a case was a live born, normally delivered at term by a 27-year-old third gravida of lower socioeconomic status. Examination of the baby revealed caudal dysgenesis having fusion of lower limbs, single leg with 1 foot and 5 toes. There was no identifiable external genitalia and anus.

Keywords: Sirenomelia; Caudal Regression Syndrome; Mermaid Syndrome; Potter's Facies.

Introduction

Sirenomelia is a rare and fatal congenital defect characterized by varying degrees of lower limb fusion, thoracolumbar spinal anomalies, sacrococcygeal agenesis, genitourinary, and anorectal atresia. The incidence of sirenomelia is 0.8-1 case/100,000 births with male to female ratio being 3:1 [2]. There is a strong association with maternal diabetes where relative risk is 1:200-250 and up to 22% of fetuses with this anomaly will have mothers with diabetes [3].

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A 27-year-old unbooked G3P1L1A1 at 39 weeks 5 days of gestational age with previous one live vaginal birth and one first trimester spontaneous abortion was admitted in the labor room with pain in the abdomen. She had no history of prior antenatal care and belonged to a tribal community with lower socioeconomic status. She was otherwise healthy with no known history of genetic or congenital anomaly in her family.

On examination, she was observed to be in the second stage of labor with cephalic presentation and regular fetal heart rate. She delivered a term 2.5 kg baby with multiple congenital anomalies. The Apgar score was 3 at 1' and 0 at 5 min. The baby died within 30 min postbirth in spite of resuscitation attempts by neonatologist. On physical examination, the infant showed narrow chest, bilateral hypoplastic thumb, fused lower limbs with a single foot and 5 toes, absent external genitalia, imperforate anus and umbilical cord with single umbilical artery There were also prominent epicanthal folds, hypertelorism, downward curved nose, receding chin, low-set soft dysplastic ears and small slit-like mouth suggestive of Potter's facies Autopsy was declined by the parents. Intrapartum and the postpartum period of mother was uneventful.

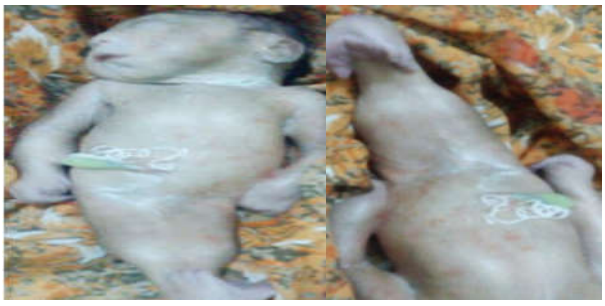

Fig. 1 & 2: Photograph of the baby showing fusion of lower limbs, hypoplastic thumb, absent external genitalia and features of Potter's facies)

Discussion

Anomalies observed in sirenomelia are described as the most severe form of caudal regression syndrome. Fusion of the lower extremities, presence of single umbilical and persistent vitelline artery are major features of sirenomelia [4,5].

Although the primary molecular defect resulting in sirenomelia remains unclear, two main pathogenic hypotheses namely the vascular steal hypothesis and the defective blastogenesis hypothesis are proposed. According to vascular steal hypothesis, fusion of the limbs results from a deficient blood flow and nutrient supply to the caudal mesoderm, which in turn results in agenesis of midline structures and subsequent abnormal approximation of both lower limb fields. However in defective blastogenesis hypothesis, the primary defect in development of caudal mesoderm is attributed to a teratogenic event during the gastrulation stage [6]. Such defect interferes with the formation of notochord, resulting in abnormal development of caudal structures. Maternal diabetes, tobacco use, retinoic acid and heavy metal exposure are possible environmental factors. In our first case, there was history of tobacco use before and during pregnancy, while in the second case the mother had overt diabetes [7].

Sirenomelia is usually fatal within a day or two of birth because of complications associated with abnormal kidney and urinary bladder development and function. In literature approximately 300 cases are reported worldwide of which 14 are from India. In most of the cases the diagnosis was performed after birth. In antenatal period, sirenomelia can be diagnosed as early as 13 weeks by using high resolution or color Doppler sonography. The condition is usually incompatible with life due visceral abnormalities especially that of renal system. Exceptional cases without renal agenesis have survived, the best example being Tiffany Yorks, a 13-year-old girl who was born with fused legs. Over the years, she has undergone numerous operations to separate her lower extremities [8].

The facial abnormality usually found in sirenomeliac infants known as Potter's facies, which includes large, low-set ears, prominent epicanthic fold, hypertelorism, flat nose and receding chin. In both of our cases, features of Potter's facies were present. When features of Potter's facies are combined with oligamnios and pulmonary hypoplasia it is known as Potter's syndrome,which was present in our second case. In our first case, the right thumb was hypoplastic, which was also previously reported. Stocker and Heifetz classified Sirenomeliac infants from Type I to Type VII according to the presence or absence of bones within the lower limb.Although we did not have radiographs to classify our case with certainty, nevertheless based on external examination, we suggest our first and second case belonged to Type IV (partially fused femurs and fused fibula) and Type I (all thigh and leg bones are present), respectively [9].

Conclusion

Sirenomelia is a rare and lethal congenital anomaly. When diagnosed antenatally, termination should be offered. However, prevention is possible and should be the goal. Regular antenatal checkup with optimum maternal blood glucose level in preconceptional period and in first trimester should be maintained to prevent this anomaly.

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A Rare Case of Twisted Ovarian Cyst in Term Gestation Patient

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Abstract

Introduction: Torsion of ovarian cyst is defined as total or partial rotation of adnexa around its vascular axis or pedicle. We may get difficulty in diagnosis in term gestation patient because of gravid uterus and symptoms of onset of labour mimicks the symptoms of twisted ovarian cyst. *Method:* Case Report 32 year G3P2L2 with 38 weeks of gestation with complain of pain in abdomen and nausea since previous day. H\O same complains twice in last month which was diagnosed as preterm labour pain and treated accordingly in other hospital. She presented herself in latent phase of labour. During monitoring, she was having continuous abdominal pain and episodes of vomiting twice. Decision of emergency L.S.C.S taken in view of fetal distress and non progress of labour. But while exploring the abdominal cavity, twisted ovarian cyst of size 10 x 8 cm in right adnexa found. Right sided Salpingoophorectomy done without untwisting the pedicle. *Conclusion:* Twisted ovarian cyst is usually symptomatic in pregnancy. but here because of Gravid Uterus, It remained undiagnosed till term gestation and required right salpingoophorectomy during L.S.C.S.

Keywords: Twisted Ovarian Cyst; Torsion; Term Gestation.

Introduction

Twisting of ovarian cyst is defined as total or partial rotation of adnexal mass around its vascular axis or pedicle [1]. Patient usually presents with acute abdominal pain. Abdominopelvic examination may reveal tender cystic mass seprate from uterus. Incidence is 2 to 5/10,000 pregnancies [2]. We may get difficulty in diagnosis because of gravid uterus and symptoms of onset of labour mimicks the symptoms of twisted ovarian cyst**.** Most commonly seen are dermoid and serous cystadenomas [3]. The exact etiology is obscure. Complications of the cysts associated with pregnancy are torsion of the cyst, rupture, infection, malignancy, impaction of cyst in pelvis causing retention of urine, obstructed labour and malpresentations of the fetus [3]. Complete torsion causes venous and lymphatic blockade leading to stasis and venous congestion, haemorrhage and necrosis.

Method

Case Report

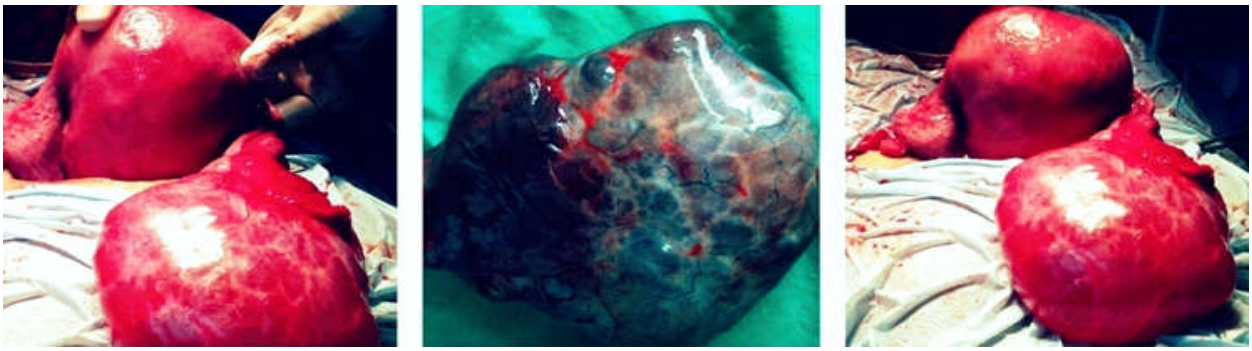
32 year G3P2L2 with 38 weeks of gestation with complain of pain in abdomen and Nausea since previous one day. H\O same complains twice in last month which was diagnosed as preterm labour pain and treated accordingly in other hospital.

Examination

previous ultrasonographic finding were within normal limit. General examination reveals she is vitally stable except pallor. On her per abdominal examination uterus was fullterm, irritable, with single live intrauterine gestation in longitudinal lie with cephalic presentation with Right occipito posterior position. as per pervaginal examination, She was in latent phase of labour. So monitoring continued.

Management

During monitoring we noticed thatshe was having continuous abdominal pain and episodes of vomiting twice, her vitals remained stable except two episodes of tachycardia lasting for few minutes. After few hours of monitoring decision of emergency L.S.C.S taken in view of fetal distress & Non progress of labour. Intraoperative period was Uneventful. But while mopping paracolic gutter and pouch of douglas, accidently we found twisted ovarian cyst of size 10 x 8 cm. in right Adnexa. Pedicle was twisted along with right fallopian tube and ovary. Right sided salpingoophorectomy done without untwisting the pedicle. Specimen sent for histopathological examination, which revealed dermoid cyst of ovary.



Discussion

Cystic teratoma, paraovarian cyst, serous cystadenoma, corpus luteal cysts, luteomas are the commonest type of ovarian tumours found during the pregnancy [3]. Differential diagnosis of this tumours are Uterine leiomyomas, appendiceal abscess, non preganant horn of bicornuate uterus, pelvic kidney, diverticular abscess, retroperitoneal tumours, ectopic pregnancy and retroverted gravid uterus [3]. During pregnancy chances of adnexal torsion raised by five times. As the clinical diagnosis in this type of cases are difficult, colour doppler is the key investigation of diagnosis. MRI is an adjuvant investigation to the doppler [4]. According to ACOG guidelines CA-125 level raises the highest level in 1st trimester and then declines further gradually. Thus CA-125 level can be helpful in 2nd & 3rd trimester for diffrentiating between malignant and benign masses [5]. Corpus luteum cyst during pregnancy is quite common, which usually regresses spontaneously by the second trimester [6]. Ovarian torsion occurs most frequently in the first trimester, occasionally in the second, and rarely in the third [7]. If we diagnose the ovarian cyst is during the first trimester, we should wait till 16 weeks, as at that time the implantation of pregnancy gets secured. Persisting tumours after 16 weeks are managed by cystectomy or ovariotomy. Ovarian tumour or cyst can be managed till 28 wks of gestation after that it is not readily accessible and may precipitate preterm labour. Ovarian cyst which ruptures, or undergoes torsion or if it shows evidence of malignancy, requires immediate surgery, irrespective of the period of gestation [8]. In our case it was an accidental diagnosis of twisted ovarian cyst in advance gestation. Retrospectively, pain & nausea-vomiting were presenting symptoms which were confused with symptoms of labour.

Conclusion

Twisted ovarian cyst is usually symptomatic in pregnancy. But here it remained undiagnosed due to hiding behind the gravid uterus till term gestation and required right salpingoophorectomy during L.S.C.S. Torsion of ovarian cyst can cause life threatening complications,which require early recognition and management accordingly. So maintenence of regular Antenatal visits and ultrasonography is utmost importance in today's Era.

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