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Case Report

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Sirenomelia following mifepristone and misoprostol intake: A case report

Abstract

Sirenomelia is a congenital birth defect that is featured as fused lower limbs, thoracolumbar anomalies, sacrococcygeal agenesis, genitourinary and anorectal atresia and Potter facies. This is a case of sirenomelia in an unsuccessful MTP. A 30-year-old came to emergency at 34 weeks with severe oligohydramnios and FGR. She had MTP pill intake in the first trimester. Her blood sugars were normal. There was no teratogen exposure. She delivered a 1.5kg male baby. In the infant there was fused lower limb, malformed genitalia with absent anogenital opening, rudimentary tail, excessive lanugo, short neck, potter's facies with prominent epicanthal folds, downward flat nose, receding chin, low set soft dysplastic ears and small slit-like mouth; suggestive of sirenomelia. Maternal diabetes is the most established etiological factor. Cocaine abuse, tobacco, misoprostol, trimethoprim, phenobarbital and carbamazepine use have reported cases of sirenomelia. Our case had MTP pill intake. Unregulated over-the-counter intake of MTP drugs should be discouraged.

Keywords: oligohydramnios, sacrococcygeal agenesis, maternal diabetes, menstrual period

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Introduction

Sirenomelia, also known as Mermaid syndrome, is a sporadic congenital birth defect which is a universally lethal condition. It has an incidence of 0.8-1 case per 100,000 births with a male-to-female ratio of 3:1.[1] Approximately 350 such cases have been reported all over the world with an incidence of only 14 cases reported in India.[2] It is featured by different degrees of fused lower limbs, thoracolumbar spinal anomalies, sacrococcygeal agenesis, genitourinary and anorectal atresia and Potter facies.[3] There are several proposed mechanisms for the following condition, but the most accepted hypotheses for mermaid syndrome are vascular steal phenomenon, defective blastogenesis, and mechanical compression of the fetal caudal body leading to defective lower extremities.[4] The strongest association of sirenomelia is seen with maternal diabetes mellitus with a relative risk of 1:200-250; where approximately 22% of mermaid babies are born to a mother with diabetes.[5] Other rare etiopathological factors have been identified such as exposure to teratogens antenatally like haloperidol, alcohol, cadmium and lead. Nutritional deficiencies are also one of the causative factors in some. We are reporting a case of sirenomelia in a patient with an unsuccessful medical termination of pregnancy with mifepristone and misoprostol in 1st trimester and the associated findings.

Case report

(People who participated in the study received Informed Consent). A 30yr old, fifth gravida female with one abortion and 3 live children with previous 3 caesarean sections, came to obstetrics emergency at 34 weeks of period of gestation (confirmed with last menstrual period and 1st trimester ultrasonography) with an ultrasound report suggestive of severe oligohydramnios and severe fetal growth restriction with normal doppler. She was an unbooked, registered case at a dispensary and was referred to Guru Teg Bahadur Hospital to manage the above-stated ultrasonography findings. She had history of MTP pill intake in the first trimester; over the counter given a positive urine pregnancy test without any documented ultrasound. Following mifepristone and misoprostol intake for 3 days, she started bleeding per-vaginum (only spotting) for 3 days with no history of passage of fleshy mass. She got an ultrasound done after 1 week of bleeding episode which was suggestive of a live intrauterine pregnancy. She then decided to continue with her pregnancy and got registered at a dispensary where she was prognosticated about MTP intake and its effects on the fetus. She was asked to follow up with blood investigations and a detailed anomaly scan (level II). However, the patient did not follow up and reported to the dispensary as her second antenatal visit at 34 weeks of gestation with an ultrasound suggestive of the above findings. She was severely anaemic with stable vitals. Her systemic examination was normal. Her haemoglobin was 7gm%, she had negative viral markers and normal urine and thyroid reports. Her Fasting and post-prandial blood sugars were 92mg/dl and 124mg/dl respectively and her HbA1c level was 4.2g%. There was no history of any toxin or teratogen exposure. The ultrasonography was suggestive of scanty liquor, and breech presentation at 29 weeks 3 days of period of gestation with an estimated fetal weight was 1.3kg. It was suggestive of severe fetal growth restriction and severe oligohydramnios with normal doppler. No gross anomaly was reported. She received 2 units of PRBCs because of severe anaemia and was started on injection dexamethasone for steroid cover. She underwent a caesarean section for fetal distress at 35 weeks and 2 days of period of gestation. She delivered a 1.5kg male baby. The APGAR scores were 3 and 5 at 1 and 5 minutes respectively. A paediatrician did resuscitation and the neonate was shifted to NICU for weak cry, central cyanosis, poor APGAR and multiple congenital anomalies. On physical examination of the infant, there was a fused lower limb (Figure 1), malformed genitalia where a penis without testes was present (Figure 2) with absent anogenital opening, rudimentary tail (Figure 3), excessive lanugo (Figure 4) and short neck. There was evidence of potter's facies with prominent epicanthal folds (Figure 5), downward flat nose, receding chin, low set soft dysplastic ears and small slit-like mouth. The skull was normal. On physical examination of the placenta, there was a single umbilical artery. The baby died after 1 hour of resuscitation as per protocol. An infantogram was performed (Figure 6) which was suggestive of

a fused femur, tibia and fibula with kyphoscoliosis of the fetal spine. The parents refused fetal autopsy. Hence, the baby was handed over to family.

Figure 1 Fused lower limb.

Figure 2 Malformed genitalia.

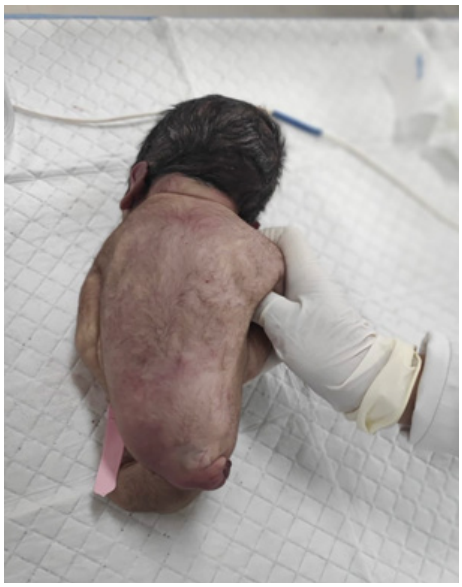

Figure 3 Rudimentary Tail.

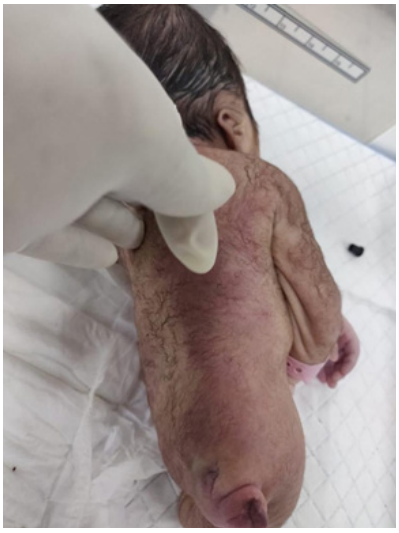

Figure 4 Excessive lanugo.

Figure 5 Potter's facies.

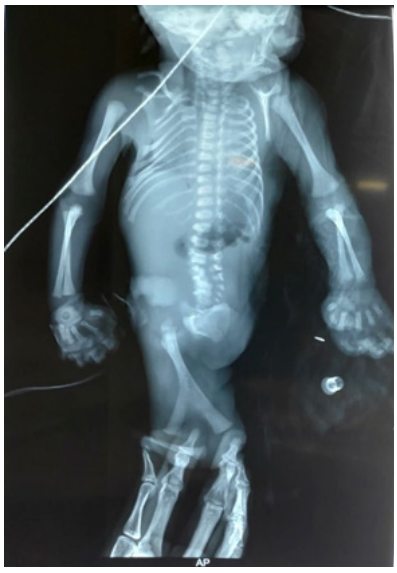

Figure 6 Infantogram of Infant.

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Discussion

Sirenomelia is a rare and lethal birth defect involving multiple organ systems. It has an analogy of the mythical creatures "sirens" or "mermaids"; hence the name.[6] In the mid-19th century, Saint-Hilaire and Forster described the three physically distinguishable variants of mermaid syndrome with different degrees of lower limb fusion i.e. (a) symelia apus: which is characterised by no feet and completely fused limbs into a single limb, in such cases only one femur and one tibia are present; our case fits into this category b) symelia unipus: characterised by one foot with 2 femurs, 2 tibia and 2 fibulae. c) symelia dipus: in this condition, two feet are present giving the appearance of fins and limbs are fused up to ankles. Our case also presented with potter's facies which can be seen in mermaid syndrome.[2] However, this classification did not include the degree of absent foot structures correlating with anomaly severity. Hence, this classification was abandoned. The current classification given by Stocker and Heifetz, has classified sirenomelia into seven subtypes based on the number of skeletal elements present in the lower limb.[7] Our case fits into type IV of this classification. However, there are reports that some fetuses do not fit into this category as well.

The exact etiology of sirenomelia is not known but several risk factors have been identified. Familial recurrence of sirenomelia and other uro- and anogenital have been reported in some cases. Maternal diabetes is the most common and established etiological factor. It is found in one in five such infants.[6] Lynch and Wright reported a case where the mother had diabetes and the infant had sirenomelia with an absent right radius and renal agenesis.[8] Castori et al.,[9] reported a diabetic mother with one child with sirenomelia and another with a VACTERL association, indicating a possible causal relationship between the two conditions.[9] Sirenomelia occurs more frequently in monozygotic twins as reported by Murphy et al.[10] Escobar reported that sirenomelia is 8 to 10% higher in twin pregnancy with approximately 100 to 150 times higher in monozygotic twins.[6] Hence, genetic counselling is suggested for future pregnancies as the risk of recurrence is 3-5%.[11] Our case was a singleton pregnancy where the patient was a non-diabetic. Teratogen exposure such as cocaine abuse, snuff tobacco, misoprostol use, trimethoprim antibiotic, phenobarbital and carbamazepine use have reported cases of sirenomelia. Mazumdar et al reported a case of sirenomelia in a patient with tobacco exposure in the first trimester of pregnancy.[12]

Every pregnancy has a background risk of 3-5% of having birth defects. Mifepristone intake in the first trimester alone does not pose a higher risk of birth defects.[13] Mifepristone when used in combination with misoprostol for medical termination of pregnancy in 1st trimester, leads to teratogenicity. Most commonly cited birth defects include equinovarus (clubfoot), cranial nerve anomalies (affecting nerves V, VI, VII, and XII), and absence of the fingers.[14] Our case had MTP pill intake in the first trimester with no other teratogen exposure.

A similar case has been reported by Bagri et al in 2016 where sirenomelia was reported in a baby born to a mother with failed first-trimester medical termination of pregnancy in a multiparous patient. No other etiological factors were identified.[15]

Conclusion

Hence, unregulated over-the-counter intake of MTP drugs (mifepristone and misoprostol) should be discouraged, and strict vigilance needs to be noted. Patients with failed medical MTP should be offered completion of the process of MTP either by surgical or other methods of termination of pregnancy, given the above-discussed chances of an anomaly in the fetus. However, if a patient is strongly willing to continue her pregnancy, detailed counselling regarding the risks to the fetus should be discussed with the couple. Routine genetic screening and detailed anomaly scans should be advised to all antenatal patients despite the presence or absence of risk factors, and timely termination should be advised in case of a lethal anomaly. The patient needs to follow the routine antenatal care with serial growth scans. Spontaneous labour is encouraged, and institutional delivery is promoted. The neonate needs to be screened by a neonatologist with timely intervention if required.

Acknowledgments

None.

Conflicts of interest

The authors have no conflicts of interest to declare.

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