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Case Report

Sirenomelia: A Case Series

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ABSTRACT

Background: Sirenomelia is a rare congenital anomaly characterized by the fusion of two lower limbs, resulting in the mermaid's tail appearance. Sirenomelia is a very rare syndrome and can be fatal to the infant. This syndrome might be sporadic or have a genetic basis. The etiology of sirenomelia is unknown but said to be multifactorial. Some maternal factors associated with sirenomelia include diabetes melitus, maternal age under 20 years and monozygote twinning.
Case report: This report presented four cases of sirenomelia that occurred during 2006-2019 at Fatemieh Hospital, a tertiary maternal-neonatal center, Hamadan, Iran. All of the pregnancies were complicated by oligohydramnios. None of the mothers were younger than 20 years, and none had diabetes mellitus. Moreover, their drug history during pregnancy was negative. All newborns had low Apgar scores and died a few minutes after birth, indicating their poor outcomes. None of the cases were diagnosed prenatally, and all newborns were firstborn infants.
Conclusion: Sirenimelia is a rare lethal condition, which requires prenatal diagnosis for abortion planning.

Keywords: Antenatal diagnosis, Congenital anomaly, Sirenomelia

Introduction

First described by Rocheus in 1542 and Palfyn in 1553 (1), Sirenomelia, also known as mermaid syndrome, is a partial or complete fusion of lower extremities associated with visceral anomalies (2). It is characterized by severe urogenital abnormalities and the presence of a singular umbilical cord blood vessel which can lead to many complications (3). Although sirenomelia has a low incidence of 1 case per every 100,000 childbirth, it is highly fatal to the infant (4, 5). The chance of live birth varies from 0.1:10,000 to 0.47:10,000 based on different reports (6). Associated malformations commonly observed in sirenomelia include renal agenesis, absent external genitalia, imperforated anus, single umbilical artery, and blind colon (7-11). Although most studies suggest that sirenomelia is sporadic, rare reports of familial cases indicate a potential genetic basis (1, 5, 7, 8). Some common sirenomelia risk factors are maternal age of under 20 years, maternal diabetes mellitus, and monozygotic twinning (2). For instance, Maternal diabetes is reported in 2% of cases (9, 12, 13). The incidence of sirenomelia is increased in monozygotic twins (7-9). Moreover, there is a higher incidence in females, with a male-to-female ratio of 1 to 3 (7, 9). Despite extensive research on sirenomelia, the etiopathogenesis of the syndrome is still controversial (9).

Case report

Case 1

A 25-year-old woman was hospitalized in March 2006 due to labor pain. According to the last menstrual period (LMP) and ultrasound reports, the pregnancy age was 32 weeks. The early third-

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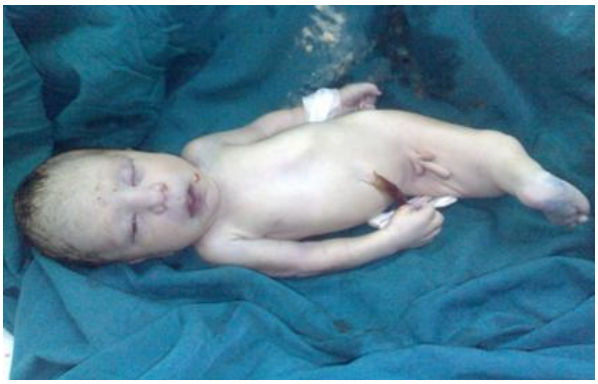

Figure 1. Case 1

trimester sonography showed only severe oligohydramnios. Several hours after admission, the baby was born with Apgar scores of 3 and 4 at the first and fifth minutes, respectively. The baby exhibited fused lower limbs from the hip downward and two feet that fused from the lateral margin with two digits in each foot (sirenomelia dipus, Figure 1). Additionally, the baby had a penis, bifid scrotum, imperforated anus, and single umbilical artery. He passed away a few minutes after birth. The X-ray findings were indicative of saccrocoxigeal agenesis and two femurs, and two tibias (Figure 2).

Case 2

A 28-year-old primiparous woman was admitted to the hospital at 38 weeks and three days of pregnancy in June 2010 because of severe oligohydramnios (AF=0) for pregnancy termination. A 2 kg baby was born with 3 and 5 Apgar scores at the first and fifth minutes, respectively. The baby had a single fused lower limb with two feet fused posteriorly and five digits

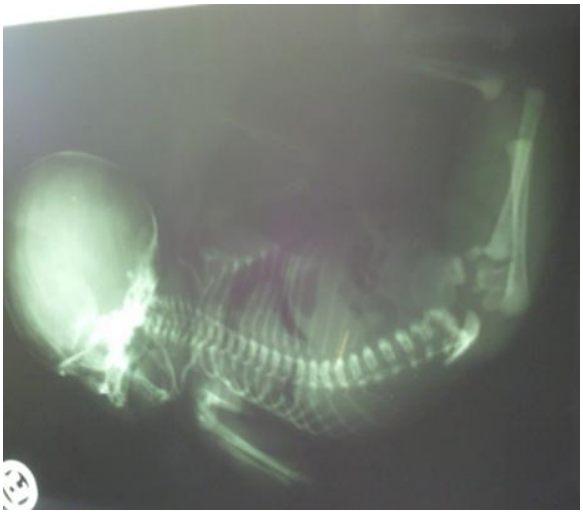

Figure 2. Whole body X-Ray of case 1

Figure 3. Case 2

in each foot, an imperforated anus, and absent external genitalia (Figure 3)

He passed away 15 minutes after birth. The X-ray findings indicated two femurs, two tibias and fibulas, small malformed metatarses, and five digits in each foot (Figure 4).

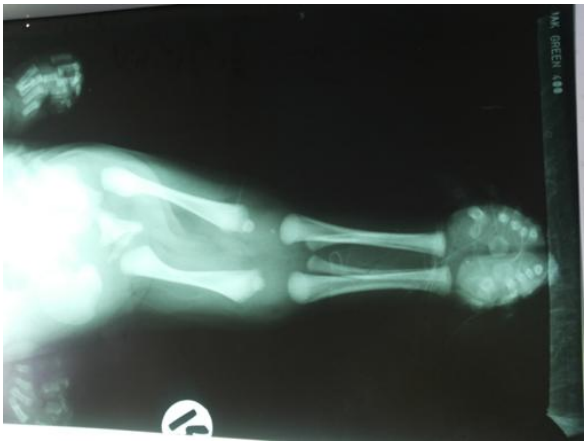

Figure 4. X-Ray of case 2

Case 3

A 22-year-primiparous woman delivered a stillborn baby at 36 weeks of pregnancy in July 2015. She was admitted to Fatemieh Hospital due to the absence of fetal movement. The baby had potter facies and sirenomelia sequence, with lower limbs fused down to the abdomen and a rudimentary digit-like tag. The baby also had an imperforated anus and absent external genitalia (Figure 5).

Case 4

24-year-old primiparous woman was admitted to the hospital with a gestational age of 32 weeks because of labor pain in August 2019. After induction, the infant was born with Apgar scores 2 and 3 in one and fifth minutes, respectively. The lower limb was fused entirely. Genitalia was absent

Figure 5. Case 3

without testis. The anus was imperforated. He expired a few minutes after birth. No X-Ray was available in this case (Figures 6,7).

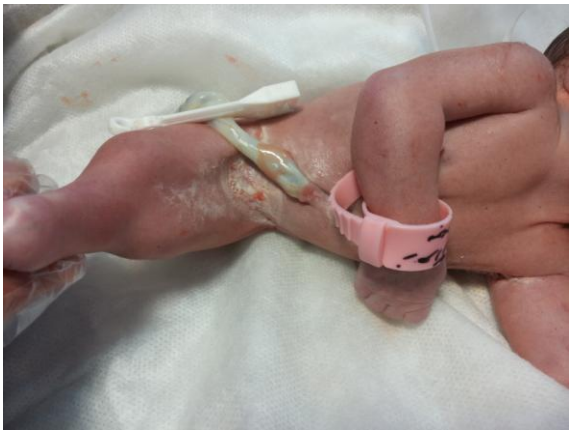

Figure 6. Case 4

All presented cases were single-tone and firstborn. Their parents were not consanguineous, and none had prenatal diagnoses. None of the mothers had diabetes, and the drug history during pregnancy was negative. Every four pregnancies were complicated by oligohydramnios.

Figure 7. Case 4

Ethical approval

This article was approved by the ethical committee of Hamadan University of Medical Sciences under the code of IR.UMSHA.REC.1402.122.

Discussion

Sirenomelia , also known as mermaid syndrome, is an extremely rare congenital developmental disorder that affects the fetal body's caudal region (14, 15). Although the fusion of the lower limbs is the most prominent characteristic of this anomaly, sirenomelia is also associated with other less visible anomalies, such as lumbosacral and pelvic bone deformities, oligohydramnios, absence of the urinary tract and external genitalia, blinding colon, single umbilical artery, anal orifice, and renal agenesis (15-17). Some of the presented cases in this report had fused lower extremities, imperforate anus, absent external genitalia, single umbilical artery, and were Small for gestational age.

Patients with caudal regression (CRS) might have similar results of this anomaly on a prenatal sonogram (18). Sirenomelia was considered a severe version of CRS in the past; however, more recent literature suggests that sirenomelia is a distinct disorder although it shares similarities with CRS (14). Fetuses with CRS typically have two umbilical arteries, two hypoplastic lower extremities, nonlethal renal findings, and imperforate or normal anus. In contrast, sirenomelic fetuses present renal agenesis or dysgenesis, single umbilical artery, fused extremities, severe oligohydramnios associated, and absence of the anus (19-21). Oligohydramnios secondary to severe renal dysplasia is widespread (10, 22). In all of the presented patients, oligohydramnios was observed as a common finding.

The incidence rate of sirenomelia is 0.8-4 per 60,000 to 100,000 pregnancies (23-25). A total of 300 sirenomelia cases have been reported, predominantly affecting males although in many cases, the sexuality is unclear due to the absence of external genitalia (10, 11, 16, 26, 27). In cases 1 and 4 in the present series, there were penis and bifid scrotum, but cases 2 and 3 had no external genitalia.

The precise etiology of sirenomelia remains unknown (5, 12, 28). Most likely, sirenomelia is multifactorial, which means that several different

factors, such as maternal diabetes mellitus, teratogenic drugs, genetic susceptibility, vascular hypoperfusion, cocaine, landfill water, male gender, and maternal age of under 20 years or over 40 years, may play a causative role for this malformation (2, 23, 25, 29-31). None of the infants in our four cases were born to diabetic mothers or mothers within the specified age range. The only identified risk for this anomaly in our cases is the male gender. Several theories have been proposed to explain the etiology of sirenomelia. The first theory suggests that an embryological insult during development leads to the abnormal fusion of the lower limbs and associated malformations. Vascular steal theory is another theory aiming to explain the etiology of sirenomelia. Sirenomelia as part of caudal regression syndrome and pressure theory, are also among other theories of sirenomelia etiology (9). The two main theories in this regard are vascular steal and defective blastogenesis theories (7, 8).

The prognosis of sirenomelia is very poor. Only about 1% of babies survive more than one week. Over 50% of the sirenomelia cases end with stillbirth, and those born alive usually die within 2 days after birth due to complications associated with a cardiac, respiratory, neurologic, genitourinary, or gastrointestinal-associated malformation (5, 32, 33). Three of these cases in this report died a few minutes after birth, and one was stillborn.

Sirenomelia can be classified into several categories. One classification, proposed by Saint-Hilaire and Forster, is based on the number of fused feet. As can be seen in Table 1, the classification by Stocker and Heifetz, provides a more detailed classification based on the number of fused bones, ranging from type I, where all bones are present, to type VII, where only a common long bone remains (8, 34, 35). Additionally, Kjaer et al. reported a less commonly used classification based on the iliac-sacral distance (34).

According to available X-ray results, the first case in this report corresponded to type III, while the second was type I. Unfortunately, the third and fourth cases had no X-rays. Sirenomelia can be easily detected in the first trimester by ultrasound (19, 26). None of the cases in this report had prenatal diagnosis despite third-trimester sonography. Oligohydramnios and fused lower limbs are crucial features that aid in diagnosing sirenomelia during the first trimester of pregnancy (15). None of the patients in this report had prenatal identification.

Table 1. Classification of sirenomelia

Type I	all thigh and leg bones are present
II	single fibula
III	absent fibulae
IV	partially fused femurs, fused fibulae
V	partially fused femurs, absent fibulae
VI	single femur, single tibia
VII	single femur, absent tibiae

Although rare, there are several reports of surviving infants with sirenomelia (2, 15, 16, 21). Six cases of surviving infants with sirenomelia have been documented until 2006 (10). However, it is important to note that all survivors need several surgical procedures. One of the key procedures involves surgically separating the conjoined legs (10, 36-38).

Conclusion

Sirenomelia is a rare, lethal syndrome. We have reported four cases of this syndrome which none of them had prenatal diagnosis. If diagnosed in early gestational age, abortion could be considered for these cases due to its mortality.

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Conflicts of interest

None is declared.

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