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CASE REPORT

SIRENOMELIA ASSOCIATED WITH HYPOPLASTIC LEFT HEART IN A NEWBORN

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ABSTRACT

Sirenomelia, also known as "mermaid malformation/syndrome," is a rare, serious congenital anomaly characterized by variable degrees of fusion of the lower limbs and associated with severe malformations of vertebral, genitourinary, cardiovascular system and single umbilical artery. The first pregnancy of a 25-year-old woman resulted in one twin born by Cesarian section at 32 weeks' gestation, who was referred to our hospital with cyanosis, a congenital anomaly and respiratory distress. On physical examination, there was no urogenital region and anal fissure and gender was indeterminate. The arms were in adduction and wrist in flexion position with four fingers on the right hand and two fingers on the left hand. There was a single lower extremity with a webbed single foot and two toes consistent with sirenomelia type IV radiologically. Abdominal ultrasonography showed urogenital system agenesis and echocardiography detected hypoplastic left heart. However, the patient died 4 hours after birth. The other twin was followed for 1 week for nutrition and respiratory support and was then discharged without any problems.

Keywords: Caudal regression syndrome (CRS); Hypoplastic left heart; Sirenomelia.

INTRODUCTION

Sirenomelia is a rare congenital anomaly, first described by Rocheus in 1542 and Palfyn in 1553 [1]. Its prevalence has been reported to be 1/600,000-1,000,000 [2]. Although the mechanism underlying sirenomelia is still unknown, maternal diabetes mellitus, genetic predisposition, vascular hypoperfusion and exposure to teratogenic agents are among probable factors [3,4].

Sirenomelia is characterized by fusion in extremities, hypoplasia or aplasia or genital system, renal agenesis and lack of bladder, single umbilical artery, vertebral defects and other skeletal system abnormalities [2]. The most common central nervous system anomalies reported in the literature are alobar holoprosocephalia and lumbar meningomyelocele. Most of the gastrointestinal system anomalies described are rectal atresia, blind end colon, imperforate anus, esophagus atresia and omphalocele. Less commonly, pulmonary defects such as lung hypoplasia, diaphragmatic hernia and cardiac defects have been reported [3-5].

Classification of syrenomelia in seven types according to Stocker and Heifetz [6] are as follows: type I, all thigh and leg bones are present; type II, single fibula; type III, absent fibula; type IV, partially fused femurs, fused fibulae; type V, partially fused femurs; type VI single femur, single tibia; type VII, single femur, absent tibia. Sirenomelia has been reported to be associated with heart defects such as truncus arteriosus, ventricular septal defect and patent ductus arteriosus [7,8]. The aim of this study was to report a case of syrenomelia type V associated with hypoplastic left heart, which has not been reported before.

CASE REPORT

A 25-year-old healthy mother's infant born at 32 weeks' gestation by Cesarian section, weighing 1040 gr, was admitted to the newborn intensive care unit because of a congenital anomaly and need for respiratory support.

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She had a dichorionic diamniotic twin. In the detailed fetal ultrasonography performed in the second trimester, oligohydramnios, single lower extremity, urinary system agenesis and single umbilical artery were detected, and in fetal echocardiography, hypoplastic left heart syndrome was reported.

Anthropometric measurements of the patient (height, weight, head circumference) were under three percentile. On physical examination, the urogenital region and anal region fissure were absent and gender was indeterminate. In the upper extremity, arms were in adduction and wrists were in flexion position. There were four fingers on the right hand and two fingers on the left hand. There was a single lower extremity, one webbed foot and two toes (Figure 1). The patient had one large umbilical artery. Radiography of the body was consistent with sirenomelia type IV (Figure 2). In bedside abdominal ultrasonography, urogenital system agenesis, and in echocardiography hypoplastic left heart were detected. However, respiratory and cardiac arrest developed and the patient did not respond to cardio-pulmonary resuscitation. The patient died 4 hours after birth. Postmortem, a 46,XY karyotype was detected. The other healthy twin was followed for 1 week for nutrition and respiratory support and was then discharged without any problems.

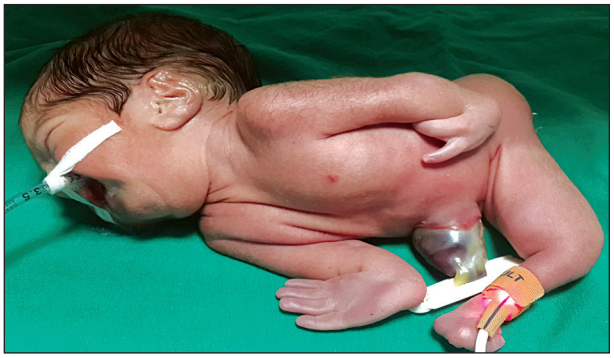

Figure 1. General appearance of the patient.

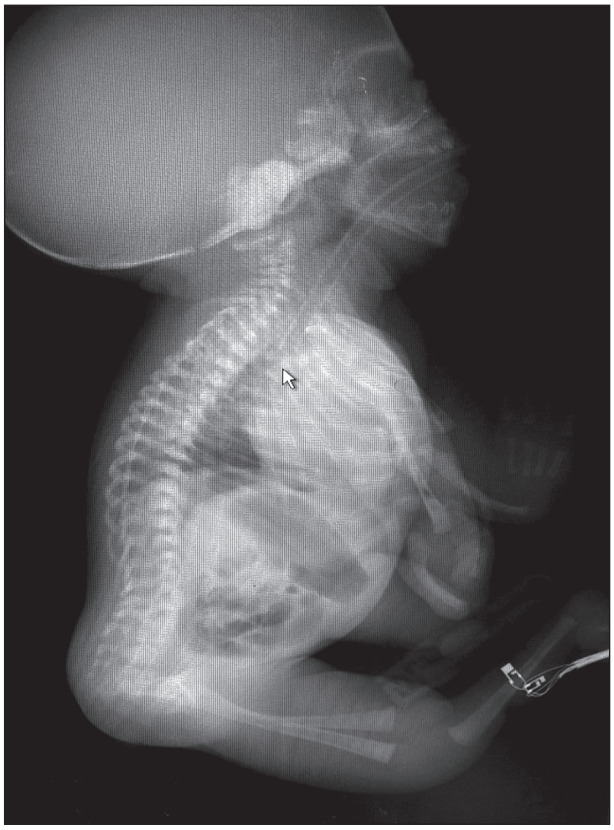

Figure 2. Whole-body X-ray of the patient.

DISCUSSION

Sirenomelia, or mermaid syndrome, is a rare syndrome characterized by serious gastrointestinal and urogenital malformations along with fusion of the lower extremities and skeletomuscular anomalies [3,4]. It is thought that sirenomelia represents the most severe form of caudal regression syndromes (CRSs). These clinical conditions form the two extremes of the spectrum derived from abnormal development of caudal mesoderm. Sirenomelia also includes extremity fusion in addition to extremity hypoplasia in CRS [4]. Although association of CRS with maternal diabetes mellitus is more frequent (22.0%), this rate has been reported to be 2.0% in sirenomelia. It is thought that free oxygen radicals, whose production increases in maternal diabetes, exerts teratogenic effect in embryonic development. In the present case, maternal diabetes mellitus and other risk factors were not present. Renal agenesis and imperforated anus occurs more commonly than CRS. As in our patient, its risk of occurrence is 100 to 150-fold higher, especially in monozygotic twins [4].

In a study by Isik Kaygusuz *et al.* [3], the presence of a single umbilical artery in sirenomelia was detected in 60.0% of cases. In this patient, a single aberrant umbilical artery was detected, which supports the vascular hypoperfusion theory in the pathogenesis of the syndrome.

Oligohydramnios, renal agenesis-hypoplasia, absence of external-internal genitalia, occur in almost all patients with sirenomelia. The earliest finding for prenatal diagnosis (PND) is oligohidramnios that can be detected as early as the 9th week of gestation [3]. In fetal ultrasonography at the second trimester, oligohidramnios, renal agenesis and a single lower extremity were detected, which was considered suggestive of sirenomelia in this patient. In postnatal abdominal ultrasonography, renal agenesis and lack of bladder were reported. Orioli *et al.* [9] reported

that gender was 46.0% ambiguous genitalia in sirenomelia, while Kırımi *et al.* [10] reported male gender at the rate of 3:1. External genitalia was not seen in this patient. Internal genital organs were also not observed in pelvic ultrasonography.

The most important mechanism underlying sironomelia is vascular steal hypothesis. According to this hypothesis, decrease in supply and blood flow of caudal mesoderm results in extremity fusion and agenesis of midline structures. Lower extremity fusion may extend from superficial tissues to bone and sirenomelia is stratified according to degree of fusion [4]. In our patient, one single lower extremity with one webbed foot and two toes were seen, which was consistent with sirenomelia type V (Figure 2). A lower extremity anomaly was reported less commonly in the literature [4,11,12]. In our case, in the upper extremities, there were four fingers on right hand and two fingers on left hand, and the arms were in adduction position.

According to the current literature, cardiac anomalies are rarely encountered in cardiac evaluation of infants with sirenomelia. In these infants, truncus arteriosus type IV is commonly seen [9]. In our patient, fetal echocardiography performed in the second trimester, hypoplastic left heart syndrome was detected and was confirmed by postnatal echocardiography. There was no strong relationship with sirenomelia and heart defects. However, Castori *et al.* [13] described a diabetic mother who had three pregnancies, which included a fetus with sirenomelia and a fetus with VACTERL association (vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies, and limb abnormalities. People diagnosed with VACTERL association typically have at least three of these characteristic features.) As is well known, heart defects are a very important component of VACTERL association. To the best of our knowledge, the association of hypoplastic heart with sirenomelia has not previously been reported in the literature.

Sirenomelia and CRSs are very similar conditions. Specific anomalies are common to both conditions, but aside from fusion of the lower extremities, an aberrant abdominal umbilical artery/persistent vitelline artery has been invoked as the chief anatomical finding that distinguishes sirenomelia from CRS [14]. Because our patient had an aberrant umbilical artery, the diagnosis was sirenomelia.

Several mechanisms have been defined to clarify sirenomelia: deficiencies in caudal mesoderm, mechanical defects resulting from lateral compression by amniotic folds and trophic defects due to a deficient blood supply in the posterior region [15]. Vascular steal hypotehesis is still the most important hypothesis in sirenomelia. According to this hypothesis, abnormal vitelline arteries would cause a reduction in blood flow to the caudal end of the embryo resulting in arrested development or aberrant formation of the lower extremities and organs. Many of these fetuses have an aberrant vasculature with the umbilical arteries connected to the old vitelline arteries (the superior mesenteric arteries). In the past, it was considered to represent the extreme form of CRS although currently, it is considered a completely different entity [15].

In conclusion, sirenomelia is a rare anomaly that is fatal due to lung hypoplasia. It may be diagnosed during the first trimester with the presence of oligohydramnios and associated anomalies. It is our suggestion that if hypoplastic left heart, which has a high mortality rate, coexists with sirenomelia, which already has a high mortality rate, this is of significance for PND and decision to terminate the pregnancy.

Declaration of Interest. The authors report no conflicts of interest. The authors alone are responsible for the content and writing of this article.

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