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BRIEF COMMUNICATION

First Trimester Diagnosis of Sirenomelia: A Case Report

Shilpa Satarkar[1]

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Abstract Sirenomelia or the mermaid syndrome is a rare and lethal congenital anomaly. It is characterized by fusion of lower extremities. It is associated with bilateral renal agenesis, anomalies of rectum, sacrum, cardiovascular system and abdominal wall. Pathognomic finding is a single umbilical artery, the 'persistent vitelline artery' which distinguishes sirenomelia from caudal regression syndrome. Sirenomelia has strong association with maternal diabetes mellitus. We report a case of sirenomelia diagnosed in first trimester. Early diagnosis was possible as protocol-based anatomic evaluation was performed. Diagnosis of sirenomelia is easier in first trimester as severe oligohydramnios in later gestation hampers the fetal evaluation.

Keywords First trimester fetal anomalies · Early diagnosis · Sirenomelia

Introduction

Sirenomelia sequence or mermaid syndrome, originally described by Rocheus in 1542 and Palfyn in 1553 is named after the mythical Greek Sirens [1, 2]. It is a very rare congenital anomaly seen in approximately 1 in 1,00,000 live births [3]. In this anomaly, the foetal legs are fused together giving the appearance of a mermaid hence it is also called as mermaid deformity. Fused lower extremities can present as a single lower limb with a deformed foot [4, 5].

This lethal congenital anomaly is frequently associated with other abnormalities including genitourinary, anorectal and cardiovascular defects [2, 5]. This syndrome has a strong association with maternal diabetes where the relative risk is 1: 200–250. Up to 22% of foetuses with this anomaly have diabetic mother [6, 7].

We report a case of Sirenomelia diagnosed in the first trimester.

Case Report

A second gravida, with uneventful first pregnancy and a normal child was referred for early morphology scan. She had regular menstrual cycles of 26–28 days. Gestational age of fetus was 11 weeks 2 days. She was nondiabetic. There was no significant medical or family history.

Her transabdominal scan and transvaginal scans were done. There was a single, live intrauterine foetus of CRL 46 mm. The singleton fetus with gestational age of 11 weeks 2 days showed single lower extremity with fixed extension at the level of the hip joint. There were two femurs in the thigh, two bones in mid leg and a single deformed foot. Mid leg level showed an echogenic twiggy area probably remnant of another leg (Fig. 1). An omphalocele was seen (Fig. 2). There was a single umbilical artery, arising directly from the aorta (Fig. 3). Further course of the aorta was very narrow and difficult to evaluate. There was reversal of 'A' wave in Ductus venosus. Nuchal translucency (NT) measured 1.3 mm and nasal bone was seen. Upper limbs were normal. Amniotic fluid was adequate.

✉ Shilpa Satarkar
shilparamesh91@gmail.com

[1] Antarang Sonography and Colour Doppler Center, Aurangabad, Maharashtra, India

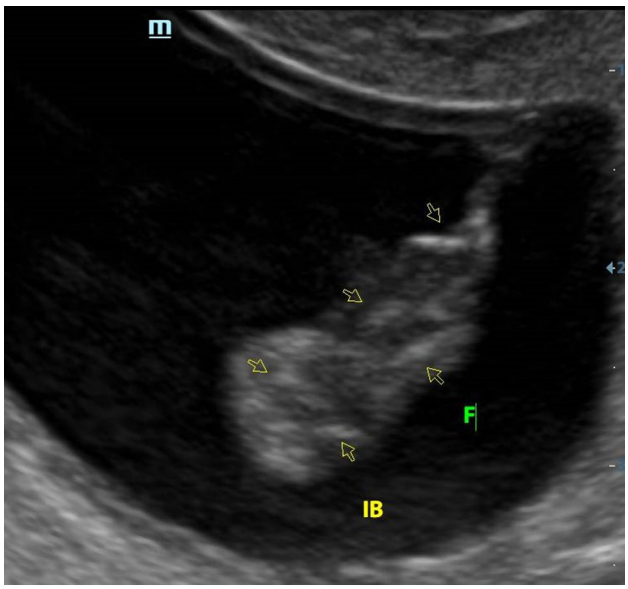

Fig. 1 Showing single lower limb with two femur in the thigh area, two bones below the knee and a single deformed foot

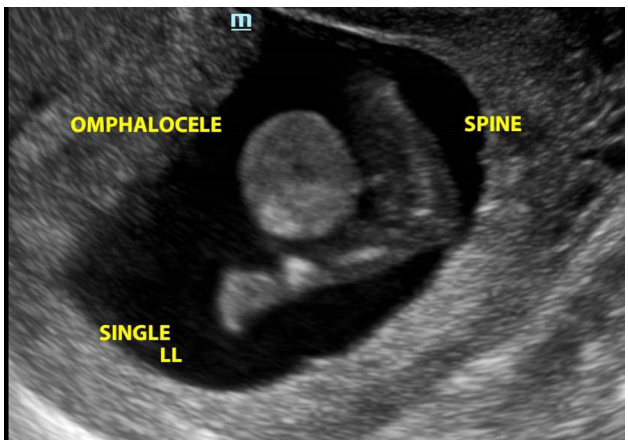

Fig. 2 Omphalocele and a single lower limb

The couple was counselled regarding poor prognosis. They were not ready for any further investigations and decided for immediate termination. They were lost to follow up.

Discussion

The ultrasound (US) diagnosis of Sirenomelia is based on the presence of fusion of the lower extremities, associated with other skeletal and lumbar spine deformities [1, 6]. In addition, bilateral renal agenesis, cardiac and abdominal wall defect could be present [6]. Diagnosis is commonly made in the second trimester while evaluating the cause of oligohydramnios [8].

In our case, diagnosis was made in the first trimester during 11–12 weeks. The diagnosis was based on the presence of single lower limb showing two femurs in the thigh. Mid leg showed two bones which could not be defined as tibia and fibula. A single deformed foot was seen. A single umbilical artery arising directly from aorta was seen in our case.

The closest differential diagnosis is Caudal Regression Syndrome. Caudal Regression Syndrome (CRS) is a rare congenital defect in the general population with a prevalence of 1–2: 100,000 in normal pregnancies [9]. Few anomalies are common to CRS and Sirenomelia. Besides fusion of the lower extremities, an aberrant single abdominal umbilical artery, the 'persistent vitelline artery' has been invoked as the main anatomic finding that distinguishes Sirenomelia from CRS [10]. In Sirenomelia, the umbilical artery is seen arising from abdominal aorta instead of Iliac Artery.

The syndrome is not associated with foetal chromosomal abnormality.

No definite etiology has been found. Two main pathogenic hypothesis, namely the vascular steal hypothesis and the defective blastogenesis hypothesis are proposed. According to vascular steal hypothesis, there is a diversion of blood flow away from the caudal portion of the embryo through the abdominal umbilical artery [1, 2, 10, 11]. This leads to 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. According to the defective blastogenesis hypothesis [10], there is the primary defect in caudal mesoderm, which is attributed to a teratogenic event during the gastrulation stage. Such defect interferes with the formation of the notochord, resulting in abnormal development of caudal structures. Maternal diabetes, use of tobacco, retinoic acid, and heavy metal exposure are possible environmental teratogenic factors [12].

Stocker and Heifetz classified Sirenomelia infants into Type I to Type VII according to the presence or absence of bones within the lower limb [13]. The sonographic diagnostic triad suggested by Raabe et al. [14] for sirenomelia/mermaid syndrome consists of (1) fused lower extremities (2) bilateral renal agenesis (3) oligohydramnios. It could not be applied in our case as the diagnosis was made in the first trimester before the onset of oligohydramnios.

Sirenomelia can be diagnosed in the first trimester. All mothers should be offered first-trimester scan with two dimensional US, colour doppler. It can be helpful in identifying a single large vitelline artery. 3D scan and MRI are helpful.

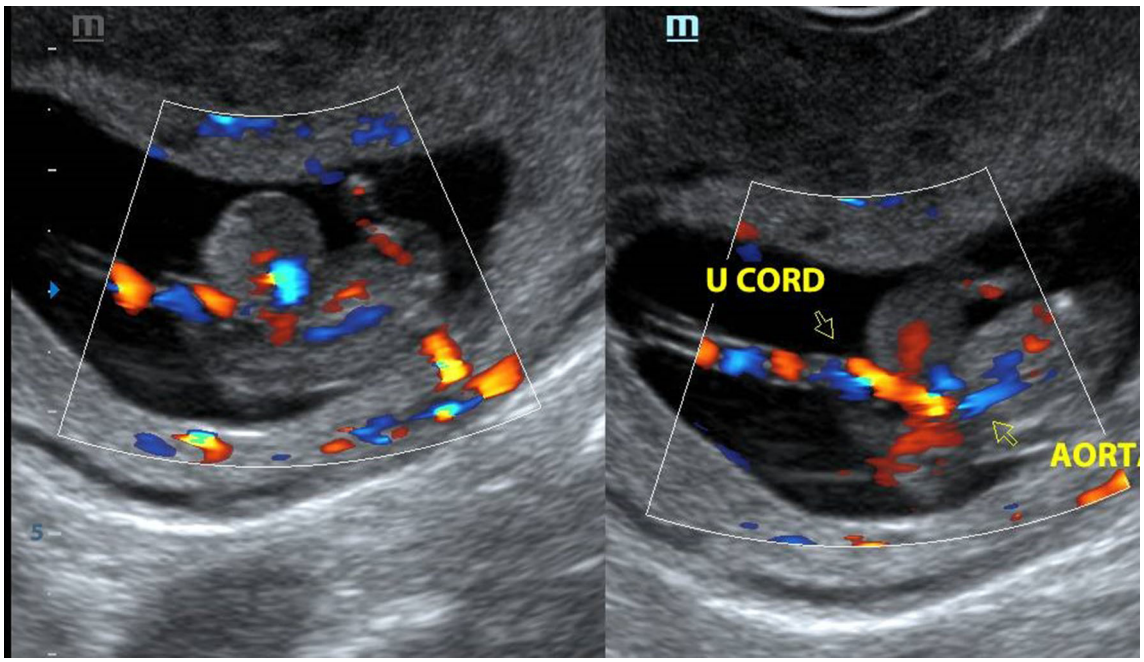

Fig. 3 Umbilical artery directly arising from Aorta

Conclusion

This case signifies the importance of protocol-based anatomical evaluation of the fetus during early morphology scan. Evaluation of limbs is easier in the first trimester, as severe oligohydramnios later in gestation hampers the views. Single umbilical artery, although not often associated with an abnormality, can also be a pointer for careful structural study. Early diagnosis of anomalies by first trimester scan should be aimed to minimize the trauma related to the termination of pregnancy at advanced gestation.

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