A rare case of Sirenomelia in Trinidad

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Abstract

Sirenomelia is a rare type of dysmelia characterized by partial or complete fusion of the legs. We present a 31-year-old $P1^{+0}$ who first presented to our antenatal clinic at 17^{+1} weeks gestational age. An ultrasonographic examination done showed a single foetus with a structurally normal head, chest, and upper limbs. The vertebrae of the lumbosacral spine appeared disorganised. The lower limbs appeared to be fused throughout its length. The ultrasonographic findings were consistent with Sirenomelia-Type 3. Although this condition is likely to have been encountered before in Trinidad and Tobago, this would be the first documented case.

Keywords: Sirenomelia, Mermaid syndrome, Antenatal ultrasonography, Trinidad & Tobago

Introduction

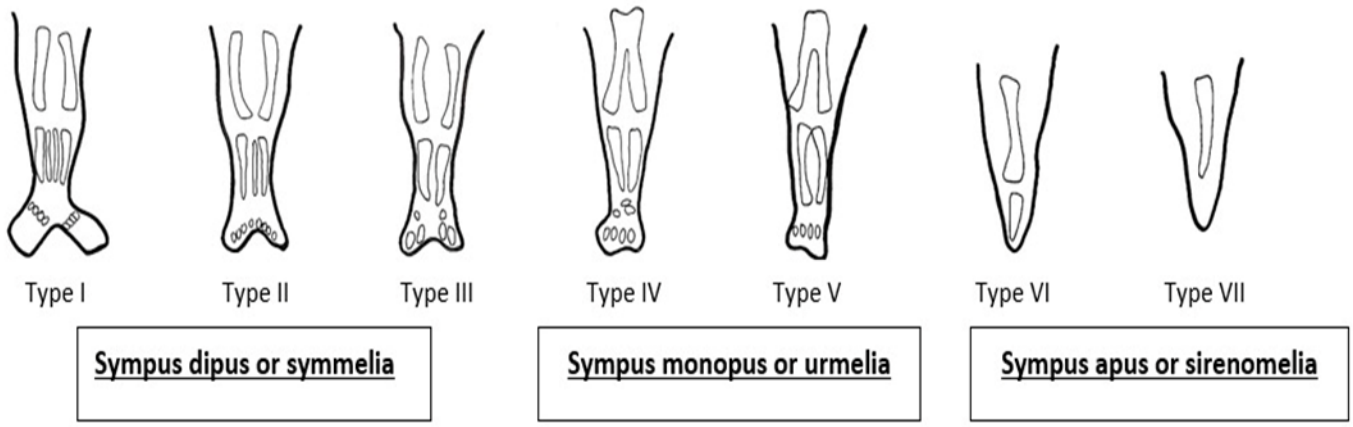
Sirenomelia originates from the ancient Greek word 'seiren' which refers to mythical creatures which were believed to be mermaids; hence it's commonly referred terminology as "Mermaid syndrome". By definition, sirenomelia is a type of dysmelia characterized by partial or complete fusion of the legs. However, it is commonly associated with a combination of lumbosacral, genitourinary, and gastrointestinal abnormalities together with a single umbilical artery. Additional findings may include an imperforate anus, absent genitalia and cardiac abnormalities.[1, 2, 3]

The incidence of Sirenomelia is 0.8 to 1 case per 1,000,000 births with a male to female ratio of 3:1.[4] In even rarer situations, sirenomelia can also occur in twin pregnancies.[4, 5, 6]

The exact cause of sirenomelia is unknown, however there have been a few hypotheses. These include environmental factors, genetic factors and the most common theory being the vascular steal theory.[1, 5, 6]

The majority of cases of sirenomelia are lethal but, there have been a few cases in which surgical management was done with survival into infancy.[1, 7] These cases, though, were dependent on the presence of at least one

Figure 1: Stocker and Heifetz classification of Sirenomelia (Source: Reference: 8)



kidney and a less severe degree of fusion of the lower limbs.

Traditionally, sirenomelia has been classified according to its extent of fusion and absence of the long bones of the lower limbs in accordance with the Stocker and Heifetz classification (Figure 1). In this case, we present the ultrasonographic and physical findings of a patient with Sirenomelia Type 3. Although, sirenomelia might have been encountered before in our setting, this may be the first documented case in Trinidad.

CASE REPORT

A 31-year-old with a parity of 1^{+0} first presented to the antenatal clinic at 17^{+1} weeks gestational age. She had a Caesarean section one year prior for foetal distress. She had no known medical conditions or allergies and she did not consume alcohol, cigarettes, or any other recreational drugs. She was taking folic acid which she started in the first trimester.

A bedside ultrasound examination at that first visit showed severe oligohydramnios with the suspicion of renal agenesis. Since Foetal Medicine services were not readily available, she was referred for a detailed anomaly scan at the Radiology Department. A second opinion was also sort from a Foetal Medicine specialist in private practice.

She returned a week later, at 18^{+1} weeks gestational age, with the ultrasonographic reports from both the Radiology Department and the private specialist (Figure 2). The reports showed a single foetus with a structurally normal brain/skull, face, chest/heart, and upper limbs. The anterior abdominal wall was intact. The vertebrae of the lumbosacral spine appeared disorganised and splayed. The kidneys were not visualised. The lower limbs appeared to be fused with no movement noticed. The femurs of both lower limbs were well formed, however, both tibias were absent. The fibulas were not fused. The feet could not be definitively identified and appeared to be fused. The foetal gender could not be ascertained. There was a single umbilical artery, severe oligohydramnios and the placenta was anterior.

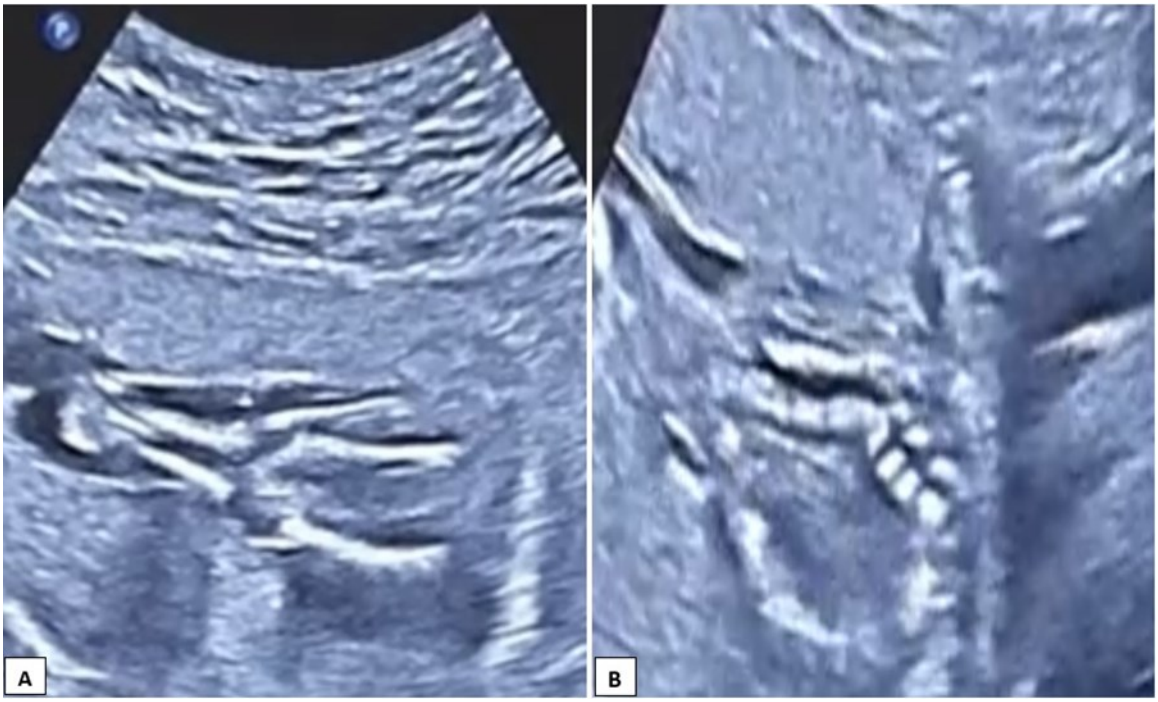
Based on the ultrasonographic findings, the patient was counselled on the lethal nature of the condition and given the option of termination of the pregnancy. She agreed to proceed with the termination which was started the next day. The delivery and post-delivery period were uneventful.

On gross examination, the foetus was noted to have a normal cranium and facial features. The upper limbs appeared normal, with normal palms and fingers. The chest and abdominal wall were normal, with no pectus excavation/carinatum or abdominal wall defects. The umbilical cord was noted to be two-vessel. The foetal external genitalia were absent, and the anus was imperforate. No vertebral defects were seen. There was complete fusion throughout the entire length of the lower limbs with posteriorly directed feet. The feet were fused and there were no toes observed (Figures 3, 4).

Discussion

Sirenomelia is a rare congenital abnormality characterized by its "mermaid like" lower limbs. It is commonly associated with a combination of other abnormalities including renal agenesis/dysgenesis, cardiac

Figure 2: Ultrasonographic images of Sirenomelia



A – Ultrasound demonstrating separate femurs but absent tibias bilaterally. Using the Stocker and Heifetz classification, this would be classified as type 3.

B – Ultrasound showing the disorganized and splayed arrangement of the lumbosacral vertebrae with abnormal curvature suggestive of a lordosis.

Figure 3: Front view of foetus with Sirenomelia demonstrating a two-vessel cord

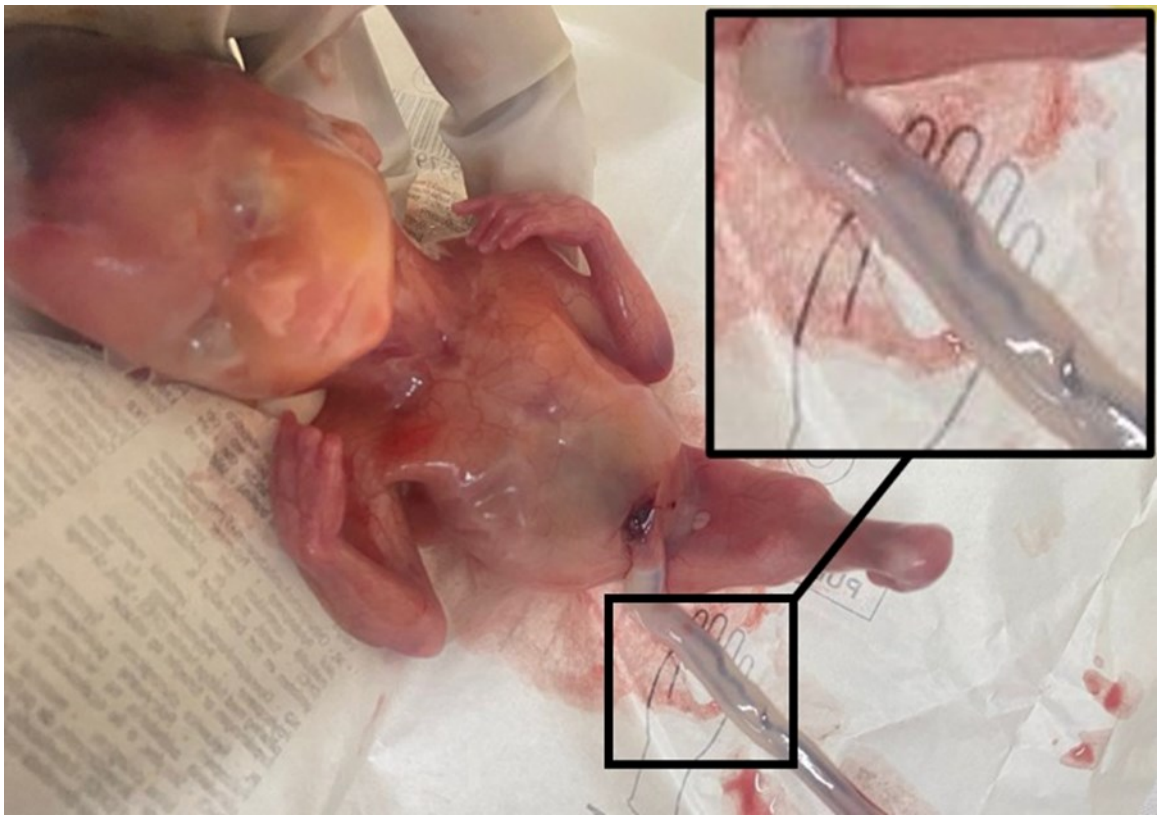
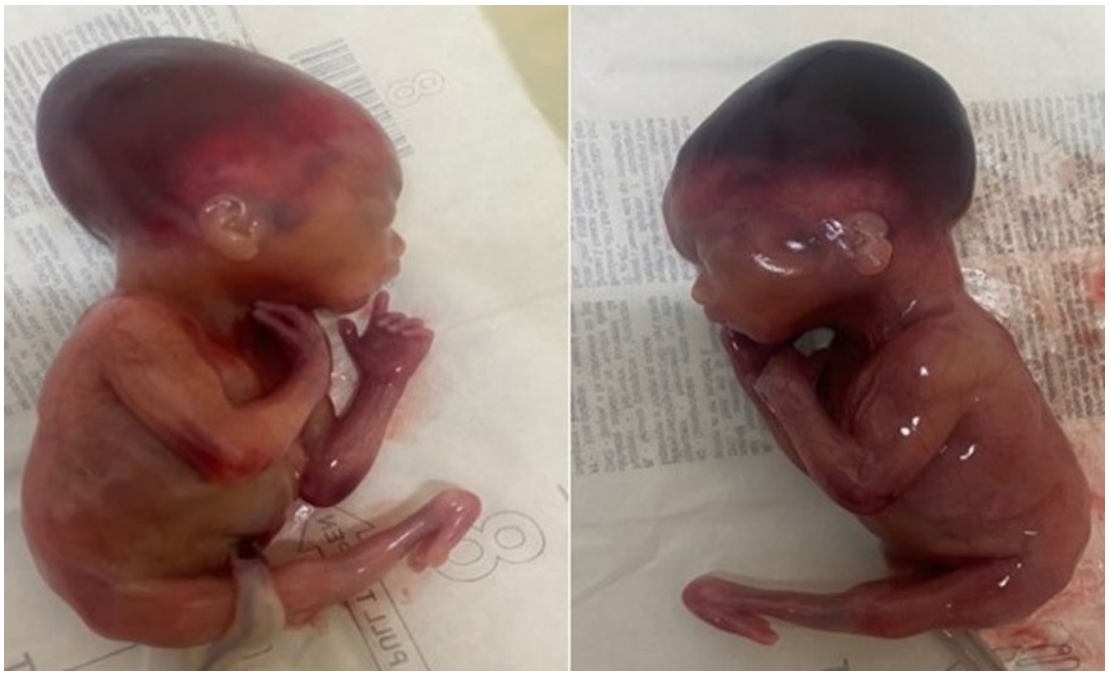


Figure 4: Side views of foetus with Sirenomelia demonstrating posteriorly facing feet



abnormalities, omphalocele, vertebral defects (particularly lumbosacral), imperforate anus, absent genitalia, and a single umbilical artery. [1, 2, 3]

The exact aetiology of sirenomelia is unknown. However, associated risk factors include extremes in maternal age, diabetes mellitus, exposure to retinoic acid, cadmium, cyclophosphamide, cocaine or lamotrigine. [5] There were no identifiable risk factors in this case and as such, may most likely be a sporadic manifestation.

A few theories exist regarding the pathogenesis of sirenomelia; however, the most accepted theory is the vascular steal theory. In this theory, there is an aberrant vessel arising from the aorta which diverts blood away from the kidneys and caudal end of the developing foetus. As a result, the lower part of the body fails to develop properly. [10]

Ultrasonographic diagnosis of sirenomelia in the first trimester is possible. In fact, it may theoretically be easier in the first trimester as severe oligohydramnios at later gestations may limit vision. [9] This case was confirmed at 18^{+1} weeks gestation since the patient presented late to the clinic, and referral for specialist services had to be arranged out of hospital.

According to Stocker and Heifetz, sirenomelia can be classified into 7 types.[8] (Figure 1) Surgical treatment can be considered for lesser degrees of fusion. Based on the ultrasonographic findings of the present case, it would be classified as Type 3. This classification, however, only considers the degree of fusion of the lower extremities and not the other commonly associated abnormalities. Consideration for surgical management would largely be dependent on the severity of the associated abnormalities. In this case, the lumbosacral spine appeared disorganised and splayed, the genitalia were not identified, there was a single umbilical artery, the kidneys were absent and there was severe oligohydramnios. Although there are documented cases of survival into infancy, this case was deemed lethal due to the non-functioning renal tissue and thus inevitable development of Potter sequence.

Obstetric practice in a low resource setting has its challenges. These include patients seeking antenatal care at a late stage or sometimes not at all, lack of high-resolution ultrasound machines, and limited expertise in detailed foetal anomaly scanning. Although this case presented at a relatively late gestation with limited ultrasonographic resources available, we were able to appropriately detect and diagnose Sirenomelia and subsequently offer suitable management.

In summary, we report a rare and first documented case of Sirenomelia in Trinidad diagnosed antenatally by ultrasonography. The condition is characterised by the appearance of 'mermaid-like' lower limbs, and is usually

associated with other congenital abnormalities. Since it is generally considered a lethal anomaly, especially those associated with renal agenesis a termination of pregnancy was undertaken in the present case.

Ethics and consent: Written consent was obtained from the patient before commencing work on this case report.

Conflict of interest : Authors declare no conflict of interest.

Author's contributions: All authors contributed to the conception and design of the work, acquisition of data, drafting the work, revising the work critically for important intellectual content, and providing final approval of the version to be published. All authors agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

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