Original Article

ISSN (O): 2347-338X; ISSN (P): 2347-3371

A Rare Case of Sirenomelia with Absent Sacrum

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Abstract

Sirenomelia is a rare lethal condition characterised by fusion of partial or complete fusion of lower limbs with male predominance. In the African context, such mermaid-like babies are called mammy -water babies, which carries an evil connotation associated with witchcraft. It is usually associated associated with diabetes milletus and tobacco usage. Risk factors are diabetes milletus, teratogenic factors, genetic factors and maternal age less than 20 years. Ultrasound is primary modality of choice of investigation in cases of sirenomelia in prenatal ultrasound. First trimester diagnosis of this disorder and induced abortion may be safest medical option. Most of the cases of sirenomelia results in still birth or die within a day or two due to congenital complications.

Keywords: Sirenomelia, Absent Sacrum

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Received: 05 September 2021 **Revised: 09 October 2021** **Accepted: 18 October 2021** **Published: 24 December 2021**

Introduction

Sirenomelia is a rare and fatal multisystemic malformation characterised by malformation of lower limbs as complete or partial fusion of lower limbs into a single lower limb, giving the appearance of a mermaid's tail.[1,2] The exact incidence is unknown, but it occurs in 1:60,000 to 1:100000 births,[3] with male to female ratio being 3:1.[4] Has a strong association with maternal diabetes where the relative risk of 1:200 to 1:250 and 22% of foetuses with this anomaly will have diabetic mothers.[5,6] It is associated with thoracolumbar spinal anomalies, sacrococcygeal agenesis, genitourinary and anorectal atresia.[7] The rarity of this case is obvious from the fact that many practioners do not come across in their carrier. Saint Hilaire and Forster classified sirenomelia based on the number of feet. The other widely used classification is the Stocker and Heifetz method, which has seven types (I-VII) and is based on the presence or absence of femur, tibia and fibula.[8,9] Ultrasound examination is the gold standard method for sirenomelia diagnosis. A first-trimester ultrasound test should be done to minimize the physical and psychological trauma related to the termination of pregnancy at longer gestational periods.

Case Report

Female with 18 weeks gestational age with no significant past medical history and low socioeconomic status came for prenatal ultrasound, ultrasound was performed. She was a known case of diabetic. On ultrasound, there was fusion of both lower limbs with absent sacrum. Both the feet were fused. Liquor was adequate for the gestational age. Bilateral kidneys were normal. Three vessels view with two umbilical arteries and one umbilical vein was seen. Four cardiac chamber was clearly seen with no defect in the cardia.

Discussions

Sirenomelia also known as mermaid syndrome is a rare congenital condition characterised by the maldevelopment of caudal portions of the body with varying degree of fusion of lower extremities. Its incidence is 1 in 60,000 to 1 in 100000 births with male predominance. Patient associated with smoking, teratogenic agents, maternal diabetes and maternal age less than 20 years are at risk.

There is no report on instances of familial recurrence of sirenomelia.[10,11] Sirenomelia as a part of caudal regression syndrome has its own pathogenesis which is maternal metabolic derangement in diabetes, but evidences have shown that sirenomelia and caudal regression are two different

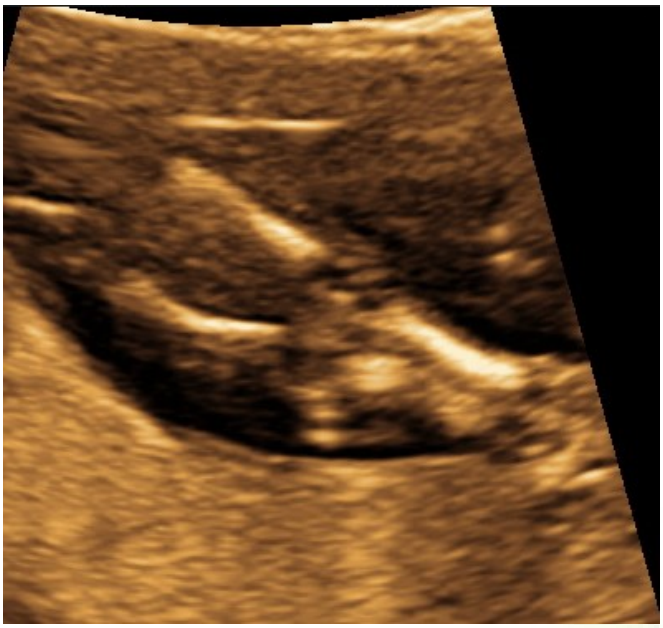


Figure 1: 18 weeks gestational age fetus clearly showing the fusion of bilateral lower limbs.

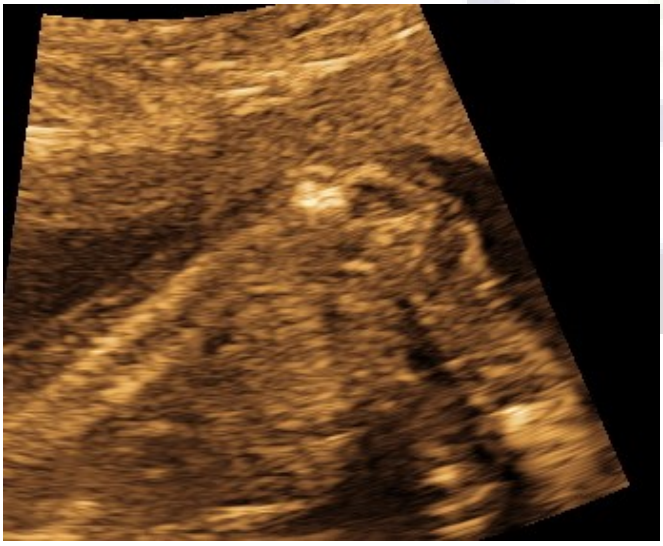


Figure 2: 18 weeks gestational age fetus showing the absence of sacrum.

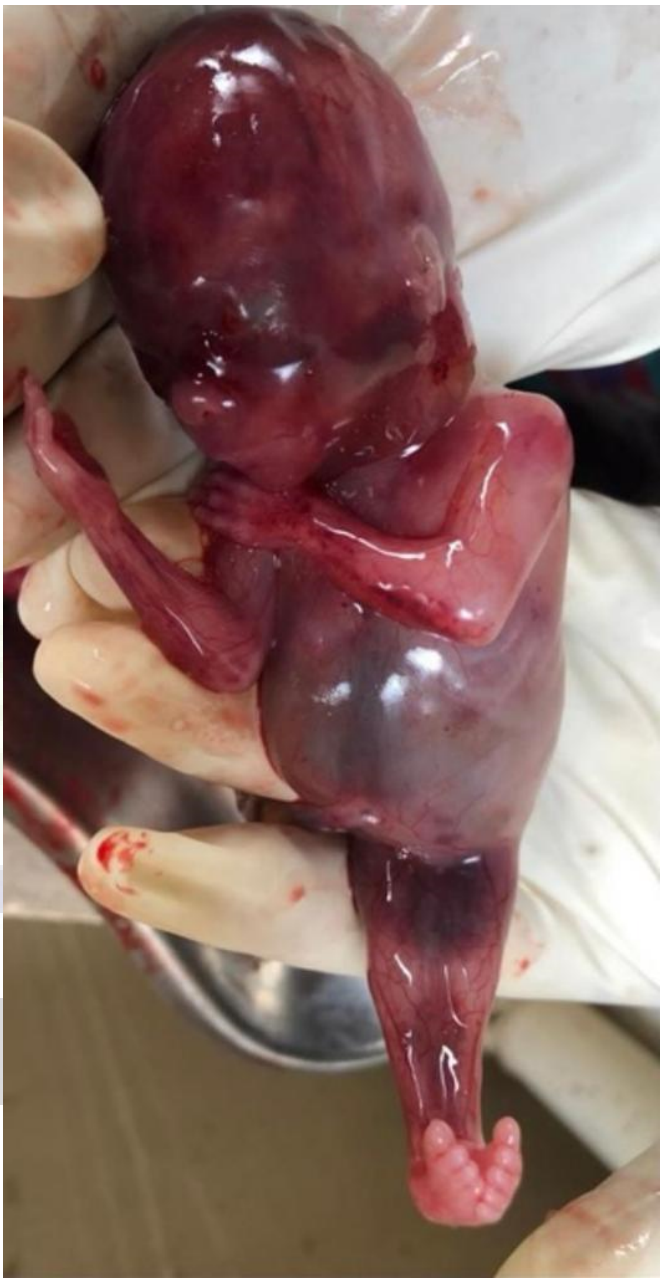


Figure 3: Photograph of the baby showing fusion of both lower limbs from hip to the feet with the absence of sacrum containing 10 toes and are fused.

entities.[12] Stevenson et al.[13]explains diversion of blood away from the caudal region of the embryo through the abdominal umbilical artery" vascular steal" has been proposed as the primary mechanism leading to sirenomelia.[14–16] Although altered oxidative metabolism from maternal diabetes may cause free oxygen radicals in the developing embryo which may be teratogenic.[17]

Saint-Hilaire and Forster classified sirenomelia into three types:[18]

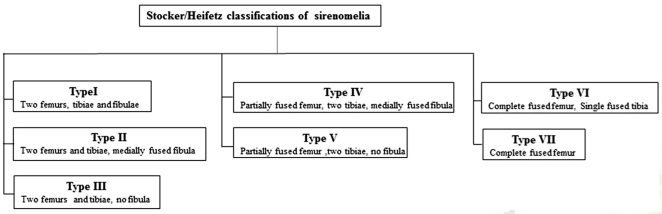
Sirenomelia Apus: There are no feet and toes, legs are conjoined fully and single femur and tibia are seen.

Sirenomelia Unipus: There is a foot but two femur, two tibia and two fibular bones seen

Sirenomelia Dipus: There are two feet and two fused legs.

Stocker and Heifetz classified sirenomelia infants from Type I to Type VII according to the presence or absence of bones in the lower limbs.

- Femur and tibia bones are formed
- Only a fibula bone seen
- No fibula bone
- The two femur and two fibula bones are conjoined imperfectly
- The two femur bones are conjoined imperfectly
- Only a femur and a tibia can be seen
- Only a femur can be seen and there is no tibia.



Most of the cases are still born or die shortly after birth. In most of the cases the diagnosis of sirenomelia was made after birth. In antenatal period, it can be diagnosed as early as 13 weeks by using high resolution or color doppler sonography. It may also be associated with malformations of musculoskeletal system (eg:sacral agenesis), gastrointestinal system (eg:omphalocle, imperforate anus), genitiurinary system (renal agenesis or hypoplasia, absent bladder). In cases of sirenomelia diagnosis can be suggested by the visualisation of a fused femur, decreased distance between two femurs or decreased/absent motility of the lower extremities.

Conclusion

Sirenomelia is one of the rarest and lethal congenital anomaly. When diagnosed antenatally, termination should be offered. Regular antenatal check-up with optimum maternal blood glucose level in preconceptional period and prevention of exposure to teratogenic agents. An early scan between 11-13 weeks can help pick up the anomaly. So, the termination of pregnancy can be planned at the earliest.

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How to cite this article: Shankar A, Jeevika MU, Solanki D. A Rare Case of Sirenomelia with Absent Sacrum. Asian J. Med. Radiol. Res. 2021;9(2):27-30.

DOI: dx.doi.org/10.47009/ajmrr.2021.9.2.7

Source of Support: Nil, **Conflict of Interest:** None declared.