Case Report

# Sirenomelia in a Nigerian: A Case Report and Review of Literature

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## Abstract

We present a case of sirenomelia delivered by a teenage mother, non-diabetic out of wedlock in a single birth. The multiple congenital anomalies involving fusion of the lower limbs, absent external genitalia, blind rectal end, absent of anal canal, single umbilical artery and embryonic mesonephric apparatus. The sex could not be determined and baby died few hours after delivery. Post mortem examination revealed multiple congenital defects. Conclusion: Rarity of single birth sirenomelia delivered by a teenage mother and emphasis on post mortem examination which revealed the gross morphological associated features that will add to the available literatures. To the best of our knowledge this is the first case to be reported in Northern Nigeria.

**Keywords:** Sirenomelia, Newborn, Post mortem

## Introduction

Mermaid has occupied the world of fantasy, fairy tale, and folklore for thousands of years. The half-human and half-fish creatures first appeared in ancient Assyria, now Syria, when the goddess Atargatis transformed herself into a Mermaid out of shame, having accidently killed her human lover. The seductive creatures have been associated with Greek mythology and referred to as sirens which mean dangerous yet beautiful creatures that lured sailors to shipwreck.[1]

In 1961, Duhamal described Sirenomelia as mermaid syndrome and the most severe form of caudal regression syndrome. It is a very rare congenital abnormality, in which the legs are fused together giving the appearance of a mermaid's tail.[2] The lesion is associated with multiple urogenital and anorectal malformations with an incidence of 0.8–1 case/100,000 live birth, with male: female ratio being 3:1.[3]

Literature review has shown about 300 cases of reported cases of Sirenomelia worldwide.[4] In Nigeria, Ugwu *et al*.[5] reported the first documented African case, in a baby of triplets. Similarly, in Lagos University Teaching Hospital, Nigeria, Odum *et al*.[6] observed congenital malformations on 22, 288 newborns with no case of Sirenomelia reported. This shows the rarity of this disorder.

To the best of our knowledge, this is the second case to be reported in Nigeria and probably in Africa.

## Case Report

A 16-year-old adolescent primigravida, got pregnant with a 25-year-old man out of wedlock, refused to attend antenatal clinics for fear of ridicule. Thought of abortion but could not bring herself for fear of exposure and denied the history of intake of drugs in the early stage of pregnancy. She is not diabetic and has no past medical or surgical history of note before delivery. She was said to be about 36 weeks pregnant when she developed sudden recurrent abdominal pain and was managed with scopolamine tablets, later that day she presented to a secondary health-care facility where she was diagnosed to be in active labor. She was managed accordingly

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and had an uneventful spontaneous vaginal delivery of a term neonate with an Apgar score of 3 in 1st min and 5 in 5th min. Attempts to resuscitate the baby failed, and the neonate died an hour later. Examination findings revealed fused lower limbs, absence of external genitalia, and absent of anal canal; other findings were essentially normal [Figure 1]. A selective nonidentity disclosure document was written and signed by the parents and doctor of the mother. This document transferred all and every right of possession of the baby. The baby was transferred to a tertiary health-care facility for postmortem studies and research.

A postmortem X-ray shows bilateral development of the skeletal bones with the absence of fibular bones-Type III [Figure 2]. A postmortem examination further revealed a single umbilical artery, blind-end colon, and embryonic mesonephric apparatus [Figure 3].

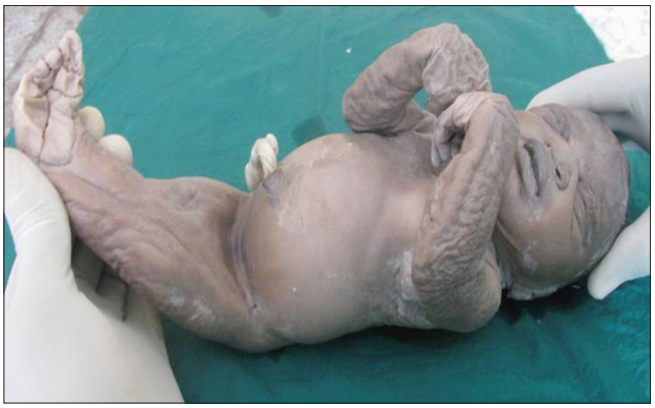

**Figure 1:** A newborn with fusion of the lower limbs

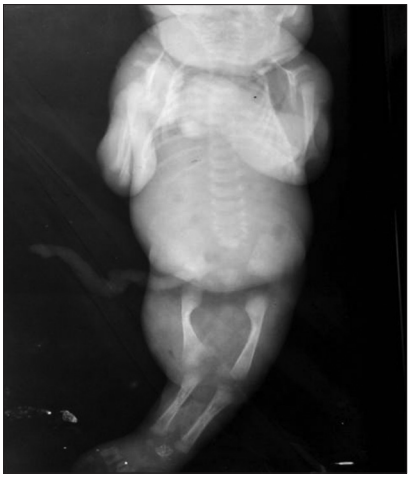

**Figure 2:** X-ray with bilateral skeletal bones, Type III

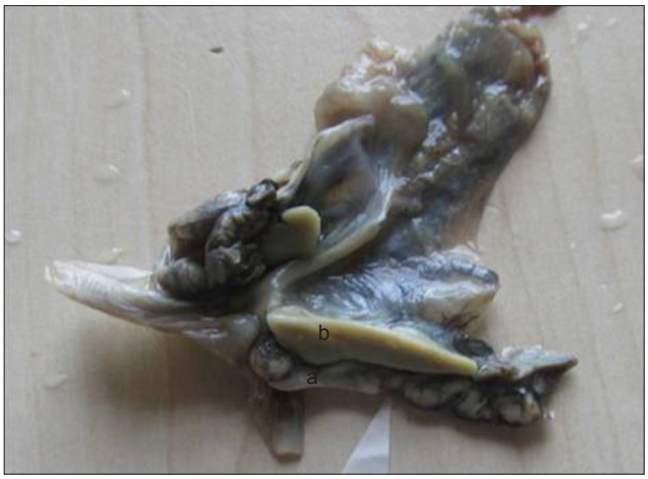


**Figure 3:** Embryonic mesonephric apparatus, mesonephric duct (a) and gonad (b)

## Discussion

Sirenomelia is rare and uncommon congenital anomalies. The cause remains unclear; however, researchers believe that both genetic and environmental factors may play a role in the development of this disorder. In some individuals, vascular steal phenomenon with the single vitelline umbilical artery diverting blood supply and nutrients from the lower body and limbs have been implicated.[7] Studies have shown Sirenomelia associated with some congenital disorders which includes the absence of spleen and gallbladder; renal agenesis; cystic malformation of the kidney; absence of bladder; urethral atresia; imperforate anus and rectum fails to developed; omphalocele; meningomyelocele; congenital heart defect; pulmonary hypoplasia; and angiomatous lumbosacral myelocystocele.[8-10]

The index case was a full-termed baby with severe congenital anomalies, and these include fusion of the lower limbs; absence of the external genitalia; absence of the anal canal, blind-end colon; embryonic mesonephric apparatus; and single umbilical artery [Figure 3].

The fusion of the lower limb is the main denominator of this disorder. The causes of this malformation remain unknown, although the discovery that it can have a genetic basis in mice represents an important step toward the understanding of its pathogenesis. Sirenomelia occurs in mice lacking Cyp26a1, an enzyme that degrades retinoic acid (RA), and in mice that develop with reduced bone morphogenetic protein (Bmp) signaling in the caudal embryonic region. The phenotypes of these mutant mice suggest that Sirenomelia in humans is associated with an excess of RA signaling and a deficit in Bmp signaling in the caudal body.

Clinical studies of Sirenomelia have given rise to two main pathogenic hypotheses. The first hypothesis, based on the aberrant abdominal and umbilical vascular pattern of affected individuals, postulates a primary vascular defect that leaves the caudal part of the embryo hypoperfused. The second hypothesis, based on the overall malformation of the caudal body, postulates a primary defect in the generation of the mesoderm.[11]

The fusion of the lower limbs may also be as postulated by the single artery theory of supplying the limbs instead

of dual arteries. Other series documented similar findings on the skeletal developmental anomalies of fusion of the lower limbs but with bilateral skeletal bones[12] similar to the index case [Figure 2]. However, other study documents only a single skeletal bone.[13] Stocker and Heifetz classified Sirenomelia into seven types based on skeletal elements in the thigh and leg. Type I is the mildest form, all bones in the two-fused limbs are present, and the fusion only affects superficial tissue. In Type VII, the most severe form, only a single bone is present with no indication of legs or feet. The index case had Type III with the absence of the fibular bones.[14]

The prenatal diagnosis of congenital disorders is very remote in most of the health-care facilities in developing countries. In most facilities, there is no fetal ultrasound which has a high-frequency sound wave to detect some of the defect associated with Sirenomelia, especially in the second trimester. The mother of this index case did not have any antenatal care or investigations during the pregnancy which could have been diagnosed. The fact that the mother is a teenager, concealing the pregnancy to term (due to out of wedlock) shows the fear and ignorance of the dangers of antenatal problems.

Sirenomelia patients that survive after 24 h and having functional kidney but have other anomalies can be surgically operated and managed.[15] This index case died a few hours after delivery because of nonfunctional kidneys (embryonic mesonephric apparatus). The arrest of the development was at an early gestational age between the 8$^{th}$ to 12$^{th}$ weeks [Figure 3]. However, the mother denied any medication for either termination of pregnancy or otherwise within the first trimester of pregnancy.

The availability of having postmortem X-ray and postmortem examinations, in this case, buttresses the importance of documenting the gross morphological features associated with Sirenomelia in this index case. This will add more to the available literature.

### Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

## Conclusion

We report a case of single-birth Sirenomelia delivered by a teenage mother and emphasis on postmortem examination which revealed the gross morphological-associated features that will add to the available literature. To the best of our knowledge, this is the first case to be reported in Northern Nigeria.

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Nil.

### Conflicts of interest

There are no conflicts of interest.

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