Case Report

Mermaid Syndrome in Enugu, Nigeria

Johnpaul Ejikeme Nnagbo[1], Chukwudi Cyril Dim[2], Hyginus Uzo Ezegwui[1]

[1]Department of Obstetrics and Gynaecology, University of Nigeria Teaching Hospital, [2]Institute of Maternal and Child Health, University of Nigeria, Ituku-Ozalla Campus, Enugu State Nigeria

Abstract

Mermaid syndrome is a type of gross fetal anomaly, characterized by the fusion of lower extremities, absent external genitalia, and apparently well-formed abdomen, chest, upper extremities, and head. The neonatal mortality rate due to this anomaly is high, but the disorder is relatively rare. Hence, few cases have been reported in the medical literature, and none in University of Nigeria Teaching Hospital, Ituku-Ozalla, Enugu State.

Keywords: Congenital anomaly, mermaid syndrome, sirenomelia

Introduction

Sirenomelia, also known as Mermaid syndrome, is a rare congenital anomaly which is often not compatible with life. It is characterized by the various degrees of anomalies of lower spine and lower limbs.[1] It is commonly associated with other anomalies and has been described as a spectrum of malformations summarized as caudal regression syndrome (CRS) malformation complex involving limb fusion defects, kidney agenesis, and anal atresia.[2-4] Since the first case was reported in 1542, efforts have been underway to understand and describe the exact cause of this syndrome, yet it still remains unclear and subject of continued debate in the medical literature.[5]

As a rare condition, it has been estimated to occur sporadically at a rate of 1 in 68,000–1 in 97,000.[4,6] The global prevalence of sirenomelia may be difficult to estimate, but as at 1992, up to 96 cases of sirenomelia were reported in a systematic review.[6] There appears not to be a particular continent not affected by this condition.[7,8] In Africa, few cases have been reported,[7-10] whereas only two were from Nigeria.[7,8] This may suggest significantly low occurrence or under-reporting of this case. Because of scanty data on sirenomelia in Nigeria and to add to the body of medical literature, we report this first case of Mermaid baby delivered at the University of Nigeria Teaching Hospital (UNTH) Ituku/Ozalla, Enugu.

Access this article online

Quick Response Code:

Website: www.njmonline.org

DOI: 10.4103/NJM.NJM_10_20

Case Report

The baby was delivered by an unbooked 30-year-old Nigerian woman whose husband was also a Nigerian. She was a primigravida who was referred to UNTH Ituku/Ozalla at a gestational age of 29 weeks with a diagnosis of severe oligohydramnios. She registered for antenatal care at 5 weeks gestational age at a private hospital and commenced intake of folic acid at the same time. No history of intake of herbal concoctions, alcohol, or tobacco in any form. Her antenatal tests such as packed-cell volume (34%), blood group: Rhesus D positive, genotype; AA, Venereal Disease Research Laboratory; nonreactive, HIV I and II; nonreactive were normal except hepatitis B surface antigen screening which was reactive, but never been symptomatic of viral hepatitis. Screening for diabetes done at the referral hospital at 5 weeks (fasting: 99 mg/dl, 2 hpp: 104 mg/dl) and 28 weeks (fasting: 112 mg/dl, 2 hours post-prandial: 143 mg/dl) was normal. Serial abdominopelvic ultrasound done at the referral hospital starting from 17 weeks persistently showed severe oligohydramnios. This prompted her referral to UNTH at 29 weeks.

Address for correspondence: Dr. Johnpaul Ejikeme Nnagbo, Department of Obstetrics and Gynaecology, University of Nigeria Teaching Hospital, Ituku-Ozalla, Enugu State, Nigeria.
E-mail: jesp7200@yahoo.co.uk

How to cite this article: Nnagbo JE, Dim CC, Ezegwui HU. Mermaid syndrome in Enugu, Nigeria. Niger J Med 2020;29:321-3.

Submitted: 25-Feb-2020 **Revised:** 05-Mar-2020
Accepted: 13-May-2020 **Published:** 26-Jun-2020

Examination of the mother at presentation showed a healthy-looking woman in no obvious distress. She was afebrile, anicteric, not pale, not dehydrated, and had no pedal edema. Her pulse rate was 78 beats/minute, full volume and regular, blood pressure was 130/80 mmHg, and heart sounds I and II were heard. There was no murmur. Her respiratory rate was 18 cycles/min, and chest was clinically clear. Her abdomen was full, moved with respiration and had a bulge at the lower region. No areas of tenderness. Her liver and spleen were not palpably enlarged, whereas the kidneys were not balottable. The symphysio-fundal height was 24 cm. There was no palpable uterine contraction. The fetal heart rate was 150 b/m (counted with fetal Doppler), strong and regular. An abdominopelvic ultrasound (two-dimensional) done at the presentation by two consultant radiologists showed a singleton live intrauterine fetus with severe oligohydramnios, one femur in the lower extremity; other lower limb bones absent, kidneys were absent instead multiple areas of hypoechogenicity were seen in the fetal pelvic area. The upper limbs, heart, and vertebral column of the fetus appeared normal. Other intrabdominal maternal organs were sonographically normal. Ultrasound impression was congenital anomalies of the renal system, ectopic (pelvic) kidneys to rule out renal agenesis. Liver function test was normal.

She was counseled on delivery, but she declined until at 32 weeks + 6 days when she decided that a cesarean section be done to deliver the baby. The neonatologist reviewed the mother and was responsible for resuscitating the baby after birth. Baby [Figures 1 and 2]: a live baby that lived up to 30 min despite all effort at resuscitation. The baby weighed 0.9 kg had a single lower extremity, absent feet and toes, no external genitalia or anus. The head, face, upper extremities, chest, spine, anterior abdominal wall, and umbilicus appeared grossly normal. She rejected radiological tests and autopsy of the fetus.

Discussion

Previous reports on sirenomelia (Mermaid syndrome) from different parts of the world centered on women with unintended pregnancies who lacked antenatal care.[7,8,10-13] However, the index case registered for antenatal care as early as 5 weeks gestational age, yet developed this condition. This is in spite of the fact that her clinical history, examination findings and investigations did not suggest any etiological factor. This clinical scenario simply suggests a sporadic occurrence.

The exact cause of sirenomelia (Mermaid syndrome) has remained a controversial issue in the medical literature for a long time. This has stimulated some research in this direction, resulting in the generation of hypothesis to explain its pathogenesis. The most popular among them are the "vascular steal hypothesis" and the "defective blastogenesis hypothesis."[14,15] The other proposed explanations include genetic, pressure, and teratogenic hypothesis. It has been established that none of these hypotheses can clearly explain all the malformations seen in mermaid syndrome.

The vascular steal theory suggests that there is an aberrant vessel which diverts blood from embryo's caudal extremity towards the placenta, depriving this part of the body of nutrients. This causes the lack of development in the lower half of the body, resulting in agenesis of the lower extremity, kidneys, genitalia and viscera. This aberrant vessel is commonly seen in the umbilical cord of the fetuses with sirenomelia as a single artery instead of two.[2]

The defective blastogenesis hypothesis was proposed based on the series of correlations reported by the researchers between abnormal caudal body in individuals with sirenomelia and defective development of caudal somites during gastrulation.[16] As a result of this, it was speculated that sirenomelia might be a severe form of CRS involving renal agenesis as a component.[17] However, a distinction has been made since sirenomelia usually presents with single umbilical artery of vitelline origin, lacked dorsal defects of neural tube and spine and also lacked clear association between it and maternal diabetes.[18-20] The baby presented had single umbilical artery and the mother did not have diabetes.

Experiments in mice had identified srn gene mutation and knockout of tsg1 genes to be responsible for the formation

Figure 1: Image of the mermaid baby during resuscitation immediately after birth

Figure 2: Image of the mermaid baby immediately after demise

of sirenomelia.[4,21] This is because these mutations lead to fusion of the hind limbs which is a typical feature seen in the case presented [Figure 1]. However, recently, it was suggested from experimental studies that genetic defects in retinoic acid and Bone morphogenic protein (Bmp) signaling in the caudal embryonic region is responsible for development of sirenomelia in mice.[20] This is the current hypothesis which has attempted to explain the genetic basis of sirenomelia in humans using mice. Bmp signaling is responsible for controlling gastrulation, stimulating and directing angiogenesis and vasculogenesis which occur as early as 3rd week in utero. The mother of the baby presented was infected with hepatitis B which was first diagnosed during antenatal care at about 5 weeks gestational age. However, occurrence of sirenomelia in humans has been linked to autosomal-dominant genetic condition with every single case resulting from new gene mutation or environmentally derived.[22] Other environmental insults or teratogens implicated in sirenomelia in various case reports include maternal diabetes, heavy metals and snuff.[16,20-22] However, these were not seen in the case presented.

Diagnosis of sirenomelia is usually confirmed at birth but strong clinical suspicion should be raised when there is severe oligohydramnios or anhydramnios as early as 14 weeks gestational age. The case presented was noted to have severe oligohydramnios from 19 weeks when the second ultrasound was done.[5] Diagnosis of anomalies in utero often requires a combination of high definition three-dimensional scan and experience. Late diagnosis may result in inadequate care and poorer outcome.

Management of this condition starts in utero and requires a multi-disciplinary approach.[1] It involves obstetricians, neonatologists, paediatric surgeons, cardiologists, orthopaedic and plastic surgeons, nephrologists and clinical psychologists. In fact, the specialties involved depends on the organs absent or anomaly found. Counselling should start early in preparation for what to expect. In the index case, the mother had extensive counselling, the neonatologists were involved prior to and during resuscitation after birth but baby deteriorated very fast immediately after delivery. Medical management is usually supportive with dialysis being the commonest supportive therapy. Multiple reconstructive surgeries are the commonly performed operation in order to restore near normal anatomy.[5]

The prognosis is usually very poor as most neonates die within few hours of birth even with supportive care.[4] Tiffany Yorks of Florida was the longest-surviving sirenomelia documented; born in May 1988 and died in February 2016.[5] The case presented died within the few minutes of birth despite the efforts to resuscitate.

Conclusion

Considering that sirenomelia is a very rare condition and a disease of theories, proper report of cases will help to increase the medical literature on this subject.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

References

1. Crawford SE. Sirenomelia. Rare Disease Database. National Organization for Rare Disorders (NORD). Available from: https://rarediseases.org/raredis eases/sirenomelia. [Last accessed on 2019 May 10].
2. Duhamel B. From the mermaid to anal imperforation: The syndrome of caudal regression. Arch Dis Child 1961;36:152-5.
3. Källén B, Winberg J. Caudal mesoderm pattern of anomalies: From renal agenesis to sirenomelia. Teratology 1974;9:99-111.
4. Holmes BL, editor. Sirenomelia. In: Common Malformations. Oxford: Oxford University Press, USA; 2011.
5. Sirenomelia. Wikipedia. Available from: https://en.m.wikipedia.org/wiki/sirenomelia. [Last accessed on 2019 Jun 29].
6. Kallen B, Castilla EE, Lancaster PA, Mutchinick O, Knudsen LB, Martinez- Frias ML, *et al*. The Cyclops and the mermaid: An epidermiological study of two types of rare formation. J Med. Genetics 1992;29:30-5.
7. Ngadda HA, Bartholomew MR, Ahmed BU, Barnabas DY, Dahiru AM, Alex U, *et al*. Sirenomelia in a Nigerian: A case report and Literature Review. Ann Trop Path 2017;8:51-3.
8. Ugwu RO, Eneh AU, Wonodi W. Sirenomelia in a Nigerian triplet: A case report. J Med Case Rep 2011;5:426.
9. Fadhlaoui A, Khrouf M, Gaigi S, Zhioua F, Chaker A. The sirenomelia sequence: A case history. Clin Med Insights Case Rep 2010;3:41-9.
10. Morfaw FL, Nana PN. Sirenomelia in a Cameroonian woman: A case report and review of the literature. F1000Res 2012;1:6.
11. Samal SK, Rathod S. Sirenomelia: The mermaid syndrome: Report of two cases. J Nat Sci Biol Med 2015;6:264-6.
12. Venna R, Vidyavathi K, Babu Rajendra Prasad CS. Sirenomelia-Mermaid syndrome with oesophageal atresia: A rare case report J Interdiscipl Histopathol 2015;3:113-6.
13. Sahu L, Singh S, Gandhi G, Agarwal K. Sirenomelia: A case report with literature review. Int J Reprod Contracept Obstet Gynaecol 2013;2:430-2.
14. Stevenson RE, Jones KL, Phelan MC. Vascular steal: The pathogenic mechanism producing sirenomelia and associated defects of the viscera and soft tissues. Paediatrics 1986;78:451-7.
15. Garrido-Allepuz C, Haro E, González-Lamuño D, Martínez-Frías ML, Bertocchini F, Ros MA. A clinical and experimental overview of sirenomelia: Insight into the mechanisms of congenital limb malformations. Dis Model Mech 2011;4:289-99.
16. Passarge E, Lenz W. Syndrome of caudal regression in infants of diabetic mothers: Observations of further cases. Pediatrics 1966;37:672-5.
17. Stocker JT, Heifetz SA. Sirenomelia. A morphological study of 33 cases and review of the literature. Perspect Paediatr Pathol 1987;10:7-50.
18. Abu-Abed S, Dollé P, Metzger D, Wood C, MacLean G, Chambon P, *et al*. Developing with lethal RA levels: Genetic ablation of Rarg can restore the viability of mice lacking Cyp26a1. Development 2003;130:1449-59.
19. Siren {Mus Musculus (House Mouse)}. Available from: https://www.ncbi.nlm.nih.gov/gene/20812. [Last updated on 2017 Apr 08].
20. Duesterhoeft SM, Ernst LM, Siebert JR, Kapur RP. Five cases of caudal regression with an aberrant abdominal umbilical artery: Further support for a caudal regression-sirenomelia spectrum. Am J Med Genet A 2007;143A: 3175-84.
21. Taghavi MM, Jafari Naveh HR, Shariati M, Morteza Zade F. Sirenomelia (mermaid syndrome): An infant from parents who used a special form of snuff. Pak J Biol Sci 2009;12:722-5.
22. Heifetz SA. Single umbilical artery. A statistical analysis of 237 autopsy cases and review of the literature. Perspect Paediatr Pathol 1984;8:345-78.