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Clinical Case

Postnatal Diagnosis of Sirenomelia in a Tertiary Hospital in Sub-Saharan Africa : Where Do We Most Often Go Wrong?

Sirénomélie de découverte postnatale dans un hôpital tertiaire en Afrique Sub-Saharien: difficultés diagnostiques

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ABSTRACT

Sirenomelia, also known as mermaid syndrome, is a very rare fatal birth defect in which the lower limbs are fused together, giving them the appearance of a mermaid's tail. It is usually associated with abnormal kidney development and genital and rectal abnormalities hence its mortality. We report a case diagnosed postnatally in a tertiary hospital in the city of Yaounde.

RÉSUMÉ

La sirénomélie est une malformation rare, toujours fatale au cours de laquelle les deux membres inferieurs sont fusionnés, donnant au fœtus un aspect de queue de sirène. Elle est presque toujours associée à des malformations du tractus génito urinaire et du rectum qui sont à l'origine de la mortalité. nous reportons ici un cas de sirénomélie diagnostiquée à la naissance dans un hôpital tertiaire de la ville de Yaoundé.

INTRODUCTION

Sirenomelia is a very rare birth defect in which the legs are fused together, giving them the appearance of a mermaid's tail. This condition occurs in about 1 in 100,000 live births [1]. Usually fatal due to associated renal malformations [2]. Its first description was made in 1542 by Rocheus and then by Polfyr in 1553 [3]. It is most commonly associated with renal agenesis, absent or malformed external and internal genitalia, a single umbilical artery, an imperforate anus, and a blind-ended large intestine [4]. Among other anomalies reported in association, angiomatous lumbosacral myelocystocele has been reported [5]. More than half of the cases of sirenomelia result in stillbirth and those born alive usually die within a day or two of birth due to complications related to abnormal kidney development and function. Only a few patients with sirenomelia have been reported to survive beyond the neonatal period [6-8]. In Africa, few cases have been documented. We present the case of a newborn with sirenomelia discovered postnatally in a tertiary hospital in the city of Yaounde, central Cameroon.

CASE PRESENTATION

The patient was 31 years old, G3P0020, 42 weeks of amenorrhea and came to our hospital for prenatal consultation and delivery. She had 6 antenatal consultations in a district hospital and had not taken any teratogenic drugs or traditional preparations. The prenatal checkups were unremarkable except for a second trimester ultrasound which showed severe oligohydramnios associated with intrauterine growth retardation at 25 weeks 3 days, with no visible malformation, and a breech fetus. She was not diabetic and had a history of two early miscarriages in 2018 and 2019 with no etiology highlighted. In addition, there was no consanguinity with her 40-year-old spouse.

On admission, general condition was good and vital parameters were within normal limits. On obstetrical examination, the uterine height was 30 cm and fetal Doppler auscultation noted a decrease in fetal heart sounds to 103 beats per minute. The fetus was in breech presentation; the vaginal exam showed a long posterior and closed cervix. An emergency cesarean section was indicated, which allowed the extraction of a newborn weighing 2200 g and measuring 49 cm in height and 32.5 cm in head circumference APGAR 7 (first minute) 5 (fifth minute) but who died 70 min later. Physical examination of the dead baby showed a typical Potter's facies and a normal upper body with fused lower limbs and two feet (Figure 1). Both feet were fused from the root to the toes, and two femurs could be distinguished on palpation, one tibia (Figure 1). The external genitalia were of the male type with an anal imperforation, examination of the umbilical cord showed one vein and a single artery.

No further investigation could be carried out because of the family's refusal.

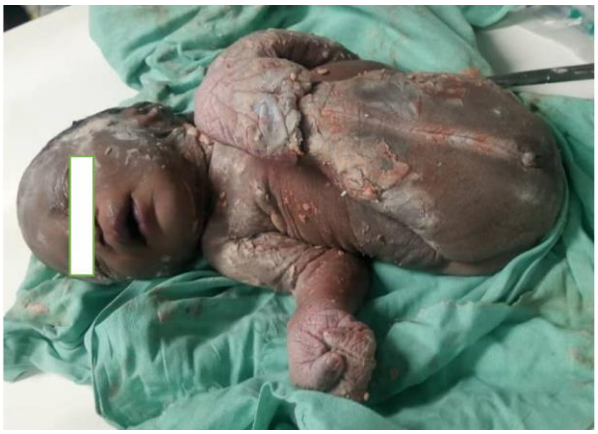

Figure 1: Mermaid fetus showing fusion of the lower limbs

DISCUSSION

Sirenomelia is a rare anomaly with high mortality, to date very little data exists on the cause of it, multiple hypotheses suggest a correlation between sirenomelia and maternal diabetes mellitus, genetic predisposition, environmental factors and the phenomenon of vascular flight with the single umbilical yolk artery diverting blood supply and nutrients from the lower body and limbs [9].

In sirenomelia, there is often an association with abnormalities of the umbilical cord blood vessels. Most babies with sirenomelia have only one umbilical artery and one umbilical vein, as was the case with our patient. The spectrum of lower limb malformations seen in babies with sirenomelia ranges from fusion of the legs into a single lower limb with only two bones (a femur and a tibia) present in the entire limb and the absence of foot structures to a simple fusion of the skin of the lower limbs along the inside with fully formed lower limb bones and fully formed feet that are fused to the ankles. In our patient we could identify two femurs and a single tibia. Although the exact type of the anomaly cannot be identified in the absence of radiographic findings, it can be classified as type II according to the classification of Stocker and Heifetz. Confusion persists as to whether sirenomelia is a severe form of VACTERL caudal regression syndrome ('vertebral defects, anorectal atresia, cardiac anomalies, tracheoesophageal fistula, renal and limb anomalies') [10]. Maternal diabetes has been associated with both caudal regression syndrome and sirenomelia, but in our case, we did not find diabetes in the mother.

The diagnosis is obvious at birth when the baby is examined, but the prenatal diagnosis can also be made in the first trimester by an ultrasound, as early as 9 weeks. In our case, the malformations were not detected, despite 3 ultrasounds done during the pregnancy. At the beginning of the pregnancy, an oligo or hydramnios related to the renal malformations makes it possible to evoke the diagnosis in the presence of malformations of the lower limbs. The diagnosis is more precise in the second trimester when there is an association between the renal malformations, the lower limbs and the oligohydramnios [12]. Nevertheless, the literature describes cases discovered in the postnatal period as in our patient. Oligohydramnios in the third trimester or at the end of the second trimester could prevent good visualization and distinction of the lower limbs [13].

The family did not give consent for X-rays, autopsy or capillary blood glucose of the mother that could allow us a better investigation. This refusal was due to the trauma caused by the sight of this newborn and his death. In Africa, malformations are often perceived as bad spells related to witchcraft and families still have difficulties in accepting autopsies.

CONCLUSION

Sirenomelia is a rare malformation with a high mortality. It exists in our context. Its diagnosis is possible in early pregnancy with a rigorous ultrasound morphology performed either by a radiologist or by an obstetrician-gynaecologist sub-specialised in antenatal diagnosis and this would allow the families to accept and prepare for the outcome of the pregnancy.

CONFLICT OF INTERESTS

The authors declare that there is no conflict of interests regarding the publication of this paper.

REFERENCES

1. Duesterhoeft SM, Ernst LM, Siebert JR, et al. 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(24):3175-84.
2. Sriram P, Venkatesh C, Srijit R, et al. Neonatal mermaid syndrome sirenomelia. J Appl Med Sci. 2010;14 (1):1-6.
3. Kampmeier OF. On sirenoform monster, with a consideration of the causation and the predominance of the male sex among them. Anat Rec. 1927; 34: 365-369
4. Sikandar R, Munim S. Sirenomelia, the mermaid syndrome: case report and a brief review of literature. J Pak Med Assoc. 2009;59 (10):721-3.
5. Raja V, Kannar V, Babu Rajendra Prasad CS. Sirenomelia-mermaid syndrome with esophageal atresia: a rare case report. J Interdiscipl Histopathol. 2015;3(3):113-6. https://doi.org/10.5455/ jihp.20150711023253.
6. Dharmraj M, Gaur S. Sirenomelia: a rare case of fetal congenital anomaly. J Med Case Rep. 2011; 5: 426.
7. Ugwu RO, Eneh AU, Wonodi W. Sirenomelia in a Nigerian triplet: a case report. J Med Case Rep. 2011. https://doi.org/10.1186/1752- 1947-5-426PMCID.
8. Khan MJ, Todase PS. Sirenomelia, the mermaid syndrome is a rare and lethal. Sirenomelia: a case report of a rare. J Clin Neonatol. 2012;1(4):221-3. https://doi.org/10.5005/Jp-journals-10053-0008.
9. Nosrati A, Naghshvar F, Torabizadeh Z, et al. Mermaid syndrome, Sirenomelia: a case report and review of literature. J Pediatr Rev. 2013;1(1):64-9.
10. Guven MA, Uzel M, Ceylaner S, et al. A prenatally diagnosed case of sirenomelia with polydactyly and vestigial tail. Genet Couns. 2008;19:419-24.
11. Stevenson RE et al. Vascular steal: the pathogenic mechanism producing sirenomelia and associated defects of the viscera and soft tissues. Pediatrics. 1986 Sep;78(3):451- 7.
12. Van Zalen-Sprock MM, Van Vugt JMG, Van der Harten JJ, Van Geijn HP. 1995. Early second trimester diagnosis of sirenomelia. Prenat Diagn 15: 171-177.
13. Sirtori M, Ghidini A, Romero R, Hobbins JC. 1989. Prenatal diagnosis of sirenomelia. J Ultrasound Med 8: 83-88.