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Sirenomelie (Mermaid Syndrome): A Case Report and Literature Review

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Abstract

Sirenomelia, or mermaid syndrome, is an extremely rare, lethal congenital malformation, occurring at a frequency of between 1.5 and 4.2 cases per 100,000 pregnancies. It is characterized by the partial or complete fusion of the lower limbs, along with malformations of the gastrointestinal and urogenital tracts. Its pathogenesis is not fully understood, with a multifactorial etiology involving genetic, teratogenic, and vascular factors. Depending on the associated malformations, death occurs in utero or within the first few days after birth. Prenatal diagnosis and imaging studies allow for reliable recognition and diagnosis. We report a case of sirenomelia diagnosed after birth, following a clinical examination of the newborn.

Keywords

Sirenomelia, Mermaid Syndrome, Congenital Malformation, Renal Agenesis, Prenatal Care Guinea

1. Introduction

A malformation is an irreversible defect in the structure of a tissue, organ, or larger part of the body resulting from an intrinsic developmental disorder. It is considered congenital if it was present at birth [1]. Caudal regression syndrome consists of sacral agenesis associated with varying deformities of the lower limbs. Sirenomelia, or mermaid syndrome, is an extremely rare and fatal congenital malformation, with an incidence of between 1.5 and 4.2 per 100,000 pregnancies [1]. It

is characterized by partial or complete fusion of the lower limbs. Its pathogenesis is not fully understood, with a multifactorial etiology involving genetic, teratological, and vascular factors [2] [3]. The presence of a single median lower limb and an aberrant abdominal umbilical artery (persistent vitelline artery) has been considered the main anatomical signs that distinguish sirenomelia from caudal regression syndrome; otherwise, they share common characteristics [4]. More than 50% of affected fetuses die in utero, and those born alive usually die within the first few days of life due to severe renal/abdominal malformations frequently associated with this syndrome [4]. Prenatal diagnosis using ultrasound and advanced imaging techniques is essential for early detection. Management is complex and requires a multidisciplinary approach involving surgery, supportive care, and rehabilitation. Although survival rates remain low due to associated complications, advances in neonatal care and reconstructive surgery are promising for improving outcomes in some cases [5] [6]. In Africa, the birth of these newborns, often referred to as "fish children", is frequently associated with mystical and religious beliefs, and the mothers of these children are often accused of witchcraft [4]. Very few cases have been reported in Africa [7] [8]. We report a case of sirenomelia diagnosed after birth at the regional hospital in Faranah, in the central-western region of the Republic of Guinea.

2. Case Report

She was a 35-year-old woman, G4 P4 with no significant family history, and already the mother of three healthy children. Throughout her pregnancy, she received no prenatal care and took no medication. There was no history of diabetes or congenital malformations in her family. Her husband, aged 50, was not related to her. At 40 weeks of amenorrhea, she presented at a local health center. Upon admission, her general condition was good and her vital signs were within normal limits. The uterine height was 32 cm. The delivery was vaginal, resulting in a newborn weighing 3.15 kg, measuring 50 cm in length, with a head circumference of 32 cm and an APGAR score of 10/10, without external genitalia, with anal imperforation and with the lower limbs fused from the base to the feet. The two feet were connected by their soles, with four toes arranged in two rows. Examination of the upper limbs revealed clenched hands (**Figure 1**). The newborn had the appearance of a "mermaid" and a diagnosis of sirenomelia was suspected. X-rays revealed the presence of a single femur, two tibias, one fibula, a single hip joint, and the absence of urinary and digestive systems (**Figure 2**). The newborn died after four days.

3. Discussion

Sirenomelia is a rare condition [9]. Its etiology remains poorly understood and is subject to debate [10]. Several pathogenic hypotheses exist. One suggests defective blastogenesis, involving impaired mesoderm development and subsequent damage to the formation of caudal structures [11]. Another hypothesis proposes that sirenomelic fetuses have a single umbilical artery of abnormal origin, derived

Figure 1. Newborn with fused lower limbs and absence of external genitalia.

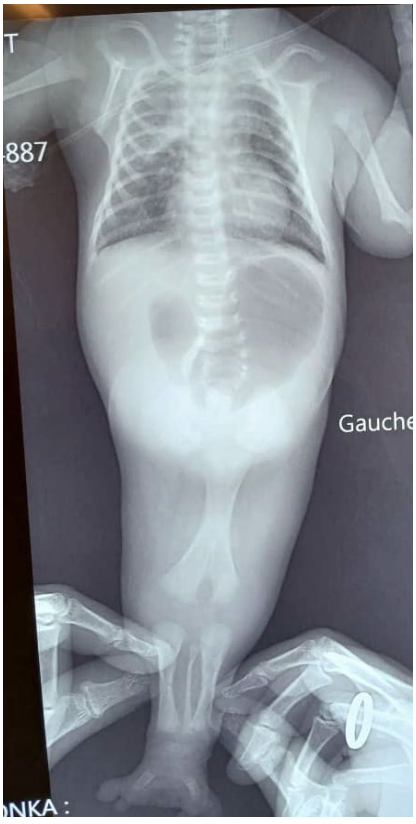

Figure 2. X-ray image showing a single fused femur, two tibias, and one fibula.

from the vitelline artery. Below the origin of this single umbilical artery, the aorta becomes severely narrowed, lacking a significant number of branches that normally supply blood to the kidneys, large intestine, and genitals. Blood is redirected from the absent or hypoplastic arteries to the single umbilical artery, which then directs blood flow to the placenta, resulting in insufficient blood circulation and nutrient supply to the lower limbs, leading to arrested development [11]. Environmental factors, particularly maternal diabetes, have been implicated [4] [10]. Indeed, 10% to 15% of reported cases involve children born to diabetic mothers, suggesting a possible role for oxidative stress, which induces an accumulation of free radicals with teratogenic effects. However, this hypothesis does not explain all cases, as only 0.5% to 3.7% of sirenomelia cases described in the literature are associated with maternal diabetes [10]. In our case, although the mother had no known history of diabetes, the lack of prenatal care does not allow us to definitively rule out gestational diabetes. From a genetic perspective, unlike in humans, it has been demonstrated that genetic alterations caused by Gain-Of-Function (GOF) mutations or Loss-Of-Function (LOF) mutations in the Bone Morphogenetic Protein (BMP) gene produce a sirenomelia-like phenotype in animal models [12]. In addition, maternal exposure to cocaine, tobacco, and alcohol, as well as exposure to teratogenic substances including air pollution, are contributing factors. Mermaid syndrome can also be caused by fetal exposure to cadmium, lithium, phenytoin, sodium valproate, carbamazepine, warfarin, methylergonovine, diethylpropion, trimethoprim, and ochratoxin (a type of fungal toxin) [13]. Due to the wide variety of malformation phenotypes, sirenomelia can be divided into several categories. Saint-Hilaire and Forster classified it as *Sympus apus* (no feet), *Sympus monopus* (one rudimentary foot), and *Sympus dipus* (two feet) [13], and the widely used classification system is the Stocker and Heifetz method, which has seven categories (I to VII) based on the presence or absence of the femur, tibia, and fibula [13] [14]. Our case resembles *Sympus monopus* with fusion of the two femurs. However, according to the Stocker and Heifetz method, it is closer to type IV due to the presence of a fused femur, two tibias, and one fibula. Regarding the case we present, the literature describes sirenomelia as a condition incompatible with life due to its association with multiple malformations and agenesis of neighboring organ systems (urinary, genital, gastrointestinal, respiratory, and neural tube defects) [14]. The diagnosis of sirenomelia should be made in the first trimester (12 - 13 weeks of gestation) using an endovaginal probe while the amniotic fluid (independent of renal function) is still normally present, facilitating visualization of the lower limbs and their movements [15] [16]. Color Doppler is essential as it can overcome the difficulty of reading due to oligohydramnios. It studies the abdominal-pelvic vascularization, specifies the level of implantation and the appearance of the abdominal aorta (complete or partial atresia) and can be used to verify the absence of renal vessels [17]. However, no prenatal assessment was performed in our context due to limited access and difficulties in the quality of prenatal care. A significant number of women in Guinea, especially in rural areas,

do not attend prenatal consultations. Ultrasound machines, which are necessary for a detailed ultrasound examination, are rare. There is a shortage of healthcare personnel specifically trained in ultrasound.

4. Conclusion

Sirenomelia is a rare and fatal congenital malformation. When diagnosed early in utero, genetic counseling is recommended for the parents, and termination of pregnancy may be considered. To our knowledge, this is the first documented case in the Republic of Guinea.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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