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Case Report

Sirenomelia (Mermaid Syndrome): First Case in Morocco and Review of the Literature

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Abstract

Sirenomelia, also known as mermaid syndrome, is a rare congenital malformation in which the lower limbs fuse together, giving the appearance of a mermaid's tail. It is frequently combined with severe urogenital and gastrointestinal anomalies. We report the case of a 30-year-old pregnant woman. During this pregnancy, sirenomelia was first diagnosed during a routine 2nd-trimester ultrasound examination, which showed fusion of the lower limbs. Neither she or any member of her family had a history of diabetes. As for other risk factors, she had no history of exposure to teratogenic agents during her pregnancy. Moreover, her marriage was not consanguineous. Sirenomelia is a rare malformative sequence that should be diagnosed as early as possible. Bilateral renal agenesis, confirmed by color Doppler, makes it a lethal condition.

Keywords: Mermaid syndrome, Sirenomelia, Caudal regression.

INTRODUCTION

The incidence of sirénomélie, a congenital anomaly, is estimated to be one case for every 60,000–70,000 pregnancies [1]. Her primary symptom is the fusion of lower members, and she is frequently linked to severe gastrointestinal and urogenital abnormalities. The primary anatomical characteristic that separates the sirénomélie from the caudale syndrome of regression is the existence of a single ombilical arter that emerges from the vitelline arter.

CASE PRESENTATION

30-year-old parturient, G2P1, with a limited pelvis, no history of diabetes and no known exposure to teratogens during pregnancy. The couple was not consanguineous.

In this 2nd pregnancy, the diagnosis of sirenomelia was made during a routine ultrasound in the 2nd trimester, then complicated by oligohydramnios. The prenatal check-up was unremarkable, with negative serologies and no gestational diabetes.

At 31 weeks of amenorrhea, she presented with hydrorhea and abdominal pain.

On admission, general condition was good and vital signs within normal limits. Uterine height was 24cm, and vaginal examination revealed an empty pelvic cavity. The patient was put on antibiotic prophylaxis and the delivery route was a caesarean section, which allowed extraction of a newborn weighing 1050g APGAR score 10/10 with absence of external genital organs and lower limbs fused from their base to the feet (Figure 1).

Figure 1: Anterior view of newborn with sirenomelia

The X-ray showed the presence of a single femur (Figure 2). The newborn had the appearance of a "mermaid".

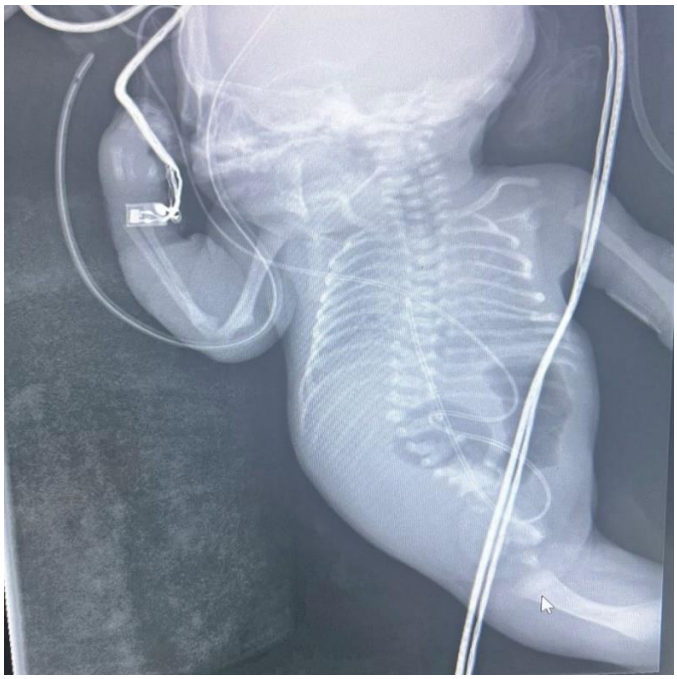

Figure 2: Radiological aspect of the sirenomelia

DISCUSSION

Sirenomelia is extremely rare, with an estimated incidence of one case per 60,000 to 70,000 pregnancies. Although the exact cause is unknown, genetic factors may play a role, as suggested by reported family cases [1].

Although the etiology of sirenomelia is not yet clear, two main pathogenic hypotheses exist. One is defective blastogenesis, which involves deficient mesoderm generation and subsequent damage to the formation of caudal structures [2]. Another hypothesis is that sirenomelia fetuses have a single umbilical artery (SUA) of abnormal origin, derived from the vitelline artery. Below the origin of the SUA, the aorta becomes severely narrow, lacking a considerable number of branches that normally supply the kidneys, large intestine and genital organs. Blood is redirected from absent or hypoplastic arteries to the SUA, which redirects blood flow to the placenta, resulting in deficient

circulation and nutrient supply to the lower limbs, leading to the arrest of their development [3].

In contrast to humans, the animal model's genetic foundation is fully known. Genetic alterations caused by gain-of-function (GOF) mutations in RA signaling or loss-of-function (LOF) mutations in bone morphogenetic protein (Bmp) have been shown to produce a sirenomelia-like phenotype in animal models. Sirenomelia in mice has been shown to be induced by genetic mutation of Cyp26a1, the main enzyme in the degradation of RA [4].

Due of the wide range of deformity phenotypes among its members, sirenomélie can be divided into multiple categories. The sirénomélie was categorized by Saint-Hilaire and Forster according to the number of feet. The other widely used classification system is the Stocker and Heifetz method, which has seven categories (I–VII) based on the presence or absence of the femur, tibia, and péroné. Less frequently used categories include the Kjaer method based on the iliaque-sacrée distance (ISD) [5].

Table 1: Stocker and Heifetz classification

Stocker and Heifetz (1987) classification based on presence of ossified structures	
Type I	Presence of two separated femurs, two tibiae, and two fibulae
Type II	Presence of two separated femurs, two tibiae, and a medially fused fibula
Type III	Presence of two separated femurs, two tibiae and no fibula
Type IV	Presence of a partially fused femur, two tibiae, and a medially fused fibula
Type V	Presence of a partially fused femur, two tibiae, and no fibula
Type VI	Presence of a complete fused femur and a single fused tibia
Type VII	Presence of a complete fused femur

Risk factors like as maternal diabetes and exposure to heavy metals for sirenomélie have been well-established in both experimental and human models, despite the fact that only 0.5 to 3.7 percent of reported cases had diabetic mothers [6].

The exposure to extrinsic factors, such as teratogenous chemicals, may cause a seirénomélie. In this regard, certain studies have discovered a connection between serométhérine and some teratogène medications, such as triméthoprime [7], phénobarbital [8], and maléate of methylergonovine [9].

However, a true sirénomélie familial recurrenc e has been reported in at least one study [10].

A prenatal diagnosis of sirenomelia is essential for an appropriate course of treatment. First- trimester ultrasound may show a fusion of lower-class members with The Doppler color is helpful in diagnosing unique umbilical artery [11].

CONCLUSION

Sirenomelia is often incompatible with life due to multiple associated anomalies. Early diagnosis enables parents to consider pregnancy management options, including medical termination of pregnancy.

Multidisciplinary management involving obstetricians, radiologists, neonatologists and geneticists is essential for comprehensive evaluation and appropriate genetic counseling.

Sirenomelia remains a diagnostic and therapeutic challenge. Research is ongoing to better understand this malformation and improve diagnostic and management strategies.

Declarations
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Consent: Informed written consent obtained from the patients.

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