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Sirenomelia, the Mermaid Syndrome in Kuwait: A case Report

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1 Abstract

Sirenomelia also called as Mermaid Syndrome, is a rare congenital malformation of uncertain etiology. It is characterized by fusion of the lower limbs and commonly associated with severe urogenital and gastrointestinal malformation. We report a case of sirenomelia occurring in a 25 year old Kuwaiti woman following premature rupture of membranes. This is the first documented case in this country.

2 Introduction

Sirenomelia is an extremely rare congenital malformative disorder, which is often fatal [1]. Its incidence is estimated at about 1 in 100,000 pregnancies [2], and cases have been reported from all ethnic groups worldwide [3]. This anomaly predominantly affects males (sex ratio 2.7:1) [4], and is frequent among one of two monozygotic twins [5]. The most prominent yet inconstant feature of this malformative disorder is the complete or partial fusion of the lower limbs into a single lower limb [3]. The resultant infant bears a resemblance to the mermaid of ancient Greek mythology [6]. The disorder has equally been referred to as symmelia, sympodia monopodia, sympus, but most commonly as the 'mermaid syndrome' since the fusion of the lower limbs gives a characteristic mermaid-like appearance [7].

The underlying visceral anomalies are usually such that the syndrome is incompatible with life [3], yet there are a number of reported cases of surviving infants with this condition in the English literature [8–12]. We report here the first documented case of sirenomelia in Kuwait, and discuss our findings in relation to the present literature and related controversies of its etiopathogenesis.

3 Case Report

Mrs. R.B, 25 years old Kuwaiti woman primigravida married for 18 months, presented to the Maternity Hospital at 31 weeks of gestation according to her last menstrual cycle with premature

rupture of membranes of 3 hours duration. This pregnancy resulted from a non-consanguineous marriage and was followed up in the private health care. She is not known to have diabetes mellitus.

On admission, the patient was hemodynamically stable and afebrile with a viable fetal heart and breech presentation. She was managed conservatively and was placed on prophylactic antibiotic. Dexamethasone course was given and she was transferred to the Antenatal Ward. 8 hours later, the patient was complaining of labor pain and digital pelvic examination revealed a fully dilated cervix with breech presentation at station +1.

Alive 1570 grams preterm baby was born by assisted breech delivery with APGAR score of 3 and 9 with endotracheal tube with the following abnormalities:

- Fused both lower limbs (Fig. 1)



Figure 1

- Ambiguous genitalia (Fig. 2)



Figure 2



Figure 3

- Cloacal anomaly (Fig. 3)
- Tracheo-esophageal fistula with atresia

The newborn was resuscitated and intubated then shifted to the Neonatal Intensive Care Unit. FISH study revealed 46XX chromosome pattern with no numerical or structural abnormalities. Skeletal survey showed 2 femurs, 2 tibia, 2 fibula with sacral segmentation defect with 13 ribs bilaterally and normal vertebrae. The baby was passing urine through a single partial opening over the sacral area. Exploratory laparotomy was done which showed distal jejunal atresia with dilated proximal bowel 10-15cm. The distal colon was atretic. The esophagus was ligated, 15cm of dilated proximal loop was resected and jejunostomy along with gastrostomy was done. Renal ultrasound showed left polycystic kidney and the right kidney was not visualized (? right ectopic kidney). The baby was discharged after 123 days to continue medical care abroad.

4 Discussion

Sirenomelia is a rare congenital anomaly characterized by partial or complete fusion of lower limbs and usually associated with other severe anomalies. The associated anomalies may include bilateral renal agenesis, complete or partial agenesis of genitourinary system, imperforate anus, absence or ambiguous external genitalia, single umbilical artery, lung hypoplasia and gastroenterology, vertebral and cardiac anomalies [13].

In our case the associated anomalies were genitourinary and gastroenterology anomalies, imperforate anus and ambiguous external genitalia.

Etiology of sirenomelia is uncertain and various theories have

been proposed to explain its origin. An embryonic insult to caudal mesoderm between 28-32 days of gestation and vascular hypo-perfusion has been proposed as possible factors [14]. Maternal diabetes has been described as an important risk factor for caudal malformations in general [3]. However, with only about 0.5–3.7% of sirenomelia cases occurring in diabetic mothers [15,18,19], the association between maternal diabetes and sirenomelia has been described as weak [20]. Our patient was not known to be diabetic. It is worth noting that although Lynch et al. [23] recognized an autosomal form of caudal dysgenesis, no chromosomal abnormalities are found in sirenomelia and it does not recur in families [16]. This was a reassuring feature for our patient and should serve as a counseling feature for mothers bearing babies with this distressing anomaly.

The etiopathogenesis of this syndrome has been subject to a lot of debate over time. Numerous theories have been proposed to explain its origin. Stevenson et al. [21] proposed the vascular steal theory. This theory suggests that there is shunting of blood via an abnormal abdominal artery arising from high up in the aorta towards the placenta. This leaves the caudal part of the embryo poorly perfused. Consequently there is hypoplasia of the vasculature distal to the artery leading to nutritional deficiency of the caudal half of the body [22]. Hence there may be complete/incomplete agenesis of the caudal structures (kidneys, sacrum, and lower portions of the digestive tract) except the gonads which are intra-abdominal. There could also be vertebral dysgenesis, lower limb atrophy and inconstant lower limb fusion [16]. The single umbilical artery in our patient favors this theory. However, Jaiyessimi et al. [20] reported a case of sirenomelia without this vitelline artery steal, indicating that factors other

Type	Characteristic
I	All Thigh and leg bones are present
II	Single fibula
III	Absent fibula
IV	Partially fused femurs,fused fibulae
V	Partially fused femurs
VI	Single femur,single tibia
VII	Single femur,absent tibia

Table 1 Classification of the Sirenomelic sequence adapted from Stocker and Heifetz2

than vitelline artery steal could be responsible for sirenomelia in humans. Like in our case. Other theories exist, but given their controversies in the literature, they are not considered here. Yet the overlap in these theories waters the debate in the scientific world as to the uniqueness or diversities of this syndrome.

Stocker and Heifetz [15] classified the sirenomelia sequence into 7 types as shown be in (Table 1). We had radiographs (fig. 4) and could therefore classify our patient into type 1. Sirenomelia car-



Figure 4

ries with it a very poor prognosis. Survival is largely dependent on the extent of visceral anomalies, especially obstructive renal failure due to renal agenesis/dysgenesis [16]. In the case of antenatal diagnosis, a voluntary termination of pregnancy is advisable in order to avoid the physical and psychological stress to parents and the family. This decision however depends on the gestational age of the pregnancy, the severity of the malformations and of course the desires of the parent s[17] and their religious beliefs. Recent reports indicate that about 50% of these infants are born alive after 8–9 months gestation [17]. However most of them die

within 5 days of life [15]. The management of sirenomelia is difficult and expensive, and the outcome is unpredictable [17]. The main therapeutic modality involves surgical and medical compensation aimed mainly at maintaining adequate renal function. Surgery to correct the anomaly and separate the fused limbs is usually not a priority as there is no guarantee of its success, and it carries with it an increased risk of compromising the life of an already delicate infant.

5 Conclusion

Sirenomelia remains a rare but peculiar syndrome. Controversies on its etiopathogenesis persist. Its prenatal diagnosis is possible albeit difficult by ultrasound. The associated visceral anomalies are usually incompatible with life. However surviving sirenomelic fetuses have been described with costly conservative management and mediocre results. The emphasis should be placed on prenatal diagnosis to ensure an optimal management that would consequently be less demanding both from a psychological and a health cost point of view.

6 References

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