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CASE REPORT

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Sirenomelia (Mermaid Syndrome): A Case Report

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Abstract

This report details the case of a neonate born at 39 weeks of gestation with dysmorphic features (Sirenomelia). After three hours of admission, the patient suffered from a cardiac arrest. Cardiopulmonary resuscitation was performed for 20 minutes, but there was no response, and the neonate died. Sirenomelia is an unusual and fatal congenital deformity, the most severe condition of caudal regression syndrome. This syndrome can cause pelvic-sacral dysplasia, genital anomalies, bilateral pelvic renal fusion with renal dysplasia, colon atresia, unilateral umbilical artery, and imperforated anus.**(International Journal of Biomedicine. 2023;13(4):377-379.)**

Keywords: sirenomelia • mermaid syndrome • neonate • X-ray

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Introduction

Sirenomelia (SML), also called the mermaid syndrome, is a rare and fatal congenital defect characterized by varying degrees of lower limb fusion, thoracolumbar spinal anomalies, sacrococcygeal agenesis, and genitourinary and anorectal atresia.(1) SML was originally described by Rocheus in 1542 and Palfyn in 1553 and named after the mythical Greek sirens.(2) The incidence of SML is 0.8 to 1 case/100,000 births, with male to female ratio being 3:1.(3) The main characteristic feature differentiating SML from caudal regression syndrome is the presence of a single umbilical artery arising at the aorta, called a "persistent vitelline artery."(4) This aberrant vessel is the basis for the hypothesis that SML results from "vascular steal" with the diversion of the blood away from the caudal embryo through the ectopic umbilical artery.(5) There are approximately 300 cases reported in the literature.(3) Except in extremely rare circumstances, SML is a fatal disease during the perinatal period, which resists any attempts at treatment.(6)

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Case Presentation

A 19-year-old primigravida arrived at the Emergency Department of Obstetrics at Palestine Hospital for Maternity and Childhood in Sana'a, Yemen. Her cervix was fully dilated and ready for vaginal delivery. During the gestational period, the mother did not seek any prenatal care and did not receive

any gestational ultrasound during pregnancy. The mother was taking supplementary folic acid that she started in the third trimester. The medical history of the mother was unremarkable, as well as the fact that the mother denied any chronic use of any medication, particularly those with known teratogenic effects. There was no family history of similar conditions, and the mother denied any consanguinity. The mother admitted to taking a herbal substance from the Qat plant, known locally as 'Al-Qatt' for recreational use during pregnancy, and she was also a smoker. The neonate was born at 39 weeks of gestation with dysmorphic features and with a gestational weight of 2.5 kg. The neonate cried immediately after birth and had an Apgar score of 5 at 1 minute and 5 at 5 minutes before becoming completely cyanosed. Resuscitation was done, intubation took place, and the neonate was shifted to the NICU. Although the patient did not resume regular spontaneous breathing, the neonate exhibited gasping breathing. As a result, intubation was performed using an endotracheal tube, and the patient was put on mechanical ventilation in synchronized intermittent mandatory ventilation mode. Following this intervention, the patient's oxygen saturation increased to 90%, and their heart rate rose to 88 bpm.

After three hours of admission, the patient suffered from a cardiac arrest. Cardiopulmonary resuscitation (CPR) was performed for 20 minutes, but there was no response, and the neonate died.

The clinical examination revealed dysmorphic features in the baby, including a wide, flat, and open anterior fontanelle, as well as a prominent flat posterior fontanelle. Additionally, during clinical examination, several other dysmorphic features were seen on the neonate, which included a wide sagittal suture, a prominent occipital bone, a depressed nasal bridge, a flat maxillary bone (displaying a 'head on chest appearance'), a short neck with folded skin, a short chest cage with widely spaced nipples, and decreased air entry bilaterally with transmitted sounds. After conducting a focused examination of each of the systems, it was noted that there were normal S1 and S2 with no added heart sounds during the cardiovascular examination. The abdomen was found to be soft and lax when the abdominal examination was conducted, and a palpable liver was detected 4 cm below the costal margin. There was a single umbilical artery and an absence of the genitalia and the anus. When examining the lower limbs, we observed a fusion of the limbs, resulting in a single limb with six digits and palpable bilateral patellae of the knees (Figures 1 and 2).

X-rays of the lower limbs displayed conjoined lower limbs with complete soft tissue fusion and partial fusion of both femora at the superior metadiaphyseal level. The two tibiae are visible, with a single fibula in the center. The feet are partially separated and exhibit a reduced number of bones (Figure 3). A skull X-ray showed a flattened occipital bone with a depressed nasal bridge. A chest X-ray showed the heart deviated to the right side with an abnormal bilateral peripheral lucency, with no definite diaphragmatic hernia (mostly related to rotation due to positioning with a suspected bilateral pneumothorax) (Figure 4). The above radiological findings are consistent with Potter syndrome type IV.[7]

Fig. 1. *Complete lower limb infusion with the absence of genitalia. A single limb with six digits and palpable bilateral patellae of the knees*

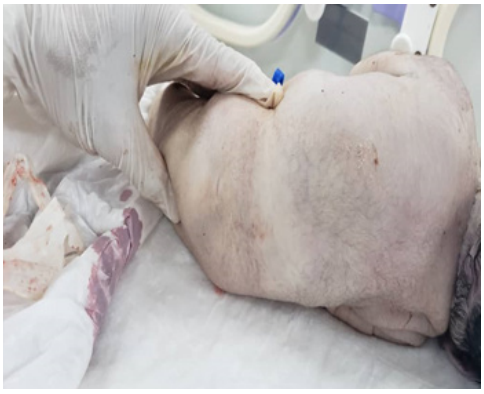

Fig. 2. *Newborn back: the absence of the anus.*

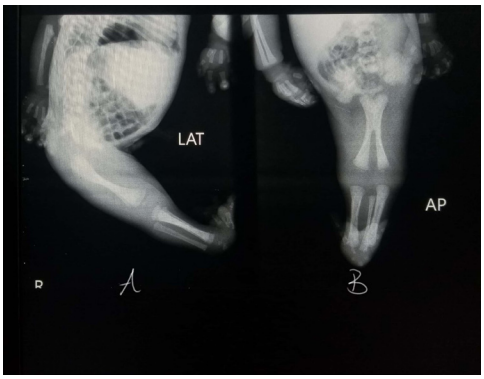

Fig. 3. *X-rays of the lower limbs: A - lateral view, B - AP view.*

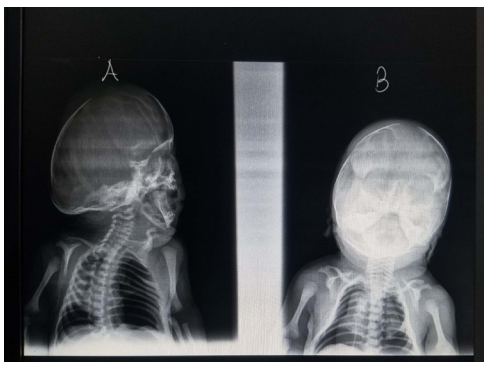

Fig. 4. *A skull & chest X-ray.*

Discussion

One of the main pathogenetic hypotheses for SML is defective blastogenesis, which is due to an abnormal development of the blastula stage during embryogenesis. Specifically, SML is characterized by a lack of mesodermal generation in the caudal region, leading to the fusion of the legs and other anomalies such as renal agenesis and gastrointestinal defects.[8]

In this case, SML was discovered after delivery due to a lack of follow-up during pregnancy. Congenital anomalies are anatomical or functional alterations that take place during fetal development. They can contribute to long-term disability, which can have serious consequences for individuals, their families, healthcare systems, and societies, in addition to being major causes of perinatal and neonatal deaths. Every year, approximately 295,000 children die during their first four weeks of life due to congenital anomalies, according to the World Health Organization.[9]

J. Serudji[10] reported, "Environmental and teratogenic factors, such as cocaine, retinoic acid, heavy metals, cyclophosphamide, and certain antibiotics, have been linked to SML in humans and animal models. In addition, nicotine, alcohol, radionuclides, diethylpropion—an appetite suppressor—organic solvents of fats, and even air pollution have been associated with SML and caudal regression syndrome, which is controversially considered as its minor form."

Additionally, Torabizadeh et al.[11] reported that maternal diabetes, genetics, irradiation exposure, and the potential teratogenic effect of vitamin A may cause SML. They also explained the association of SML with new reproductive technologies, namely with ICSI (Intra Cytoplasmic Sperm Injection).

The best method to diagnose SML is ultrasonography in the first or early second trimester. The use of transvaginal ultrasound can provide detailed information on the anatomy, while color and power Doppler ultrasound can aid in the diagnosis of a single umbilical artery. Therefore, regular ultrasound examinations are recommended to detect this congenital malformation early.[12] On the other hand, MRI is helpful in advanced cases.

SML is lethal in most cases due to pulmonary hypoplasia and renal agenesis. To prevent SML, it is important to avoid teratogenic factors and maintain normal blood glucose levels in cases of maternal diabetes. Primary prevention of SML is possible through appropriate pre-conceptional diagnosis and regular follow-up during pregnancy, especially in the first trimester.

Competing Interests

The authors declare that they have no competing interests.

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