

Sirenomelia: A case report

Swagat Mahapatra, Suruchi Ambasta

ABSTRACT

Introduction: Sirenomelia is a congenital structural anomaly characterized by abnormal development of the caudal region of the body with varying degrees of fusion of lower limbs. Most of the times, the condition is fatal for the baby. Most babies do not survive even after surgery. Fifty percent of cases are seen as stillbirths, and it is much more common in identical twins. This abnormality was initially confused with caudal regression syndrome, but later was given a new name, i.e., sirenomelia mermaid syndrome.

Case Report: We present a case of a 15-year-old boy with partially fused lower limbs. A provisional diagnosis of sirenomelia is made and detailed review and surgery was planned. The boy did not report for further treatment.

Conclusion: Very rare disorder with prevalence of 1 in 100,000 live births with a total of 300 cases reported till today in which nine are from India. The precise etiology of sirenomelia was not well understood. Many theories have been proposed but none of these is considered definitive. It is very important to diagnose this condition by ultrasonography so that termination of pregnancy can be carried out.



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CASE REPORT

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Sirenomelia: A case report

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ABSTRACT

Introduction: Sirenomelia is a congenital structural anomaly characterized by abnormal development of the caudal region of the body with varying degrees of fusion of lower limbs. Most of the times, the condition is fatal for the baby. Most babies do not survive even after surgery. Fifty percent of cases are seen as stillbirths, and it is much more common in identical twins. This abnormality was initially confused with caudal regression syndrome, but later was given a new name, i.e., sirenomelia mermaid syndrome. **Case Report:** We present a case of a 15-year-old boy with partially fused lower limbs. A provisional diagnosis of sirenomelia is made and detailed review and surgery was planned. The boy did not report for further treatment. **Conclusion:** Very rare disorder with prevalence of 1 in 100,000 live births with a total of 300 cases reported till today in which nine are from India. The precise etiology of sirenomelia was not well understood. Many theories have been proposed but none of these is considered definitive. It is very important to diagnose this condition by ultrasonography so that termination of pregnancy can be carried out.

Keywords: Sirenomelia, Caudal regression syndrome, Deformity, Congenital structural anomaly, Fusion

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INTRODUCTION

Sirenomelia is a congenital structural anomaly characterized by abnormal development of the caudal region of the body with varying degrees of fusion of lower limbs. Almost always the condition proves fatal for the baby. Most babies do not survive even after surgery. Fifty percent of cases are seen as stillbirths, and it is much more common in identical twins. After initial confusion with caudal regression syndrome, it was later given a new name—sirenomelia mermaid syndrome. Sirenomelia is characterized by complete fusion of bilateral lower limbs, along with renal agenesis, gastrointestinal defects and absent external genitalia. An important finding which differentiates these two entities is the presence of single umbilical and persistent vitelline artery.

CASE REPORT

A 15-year-old boy presented to our outpatient department with deformity of both lower limbs with partial fusion of lower legs since birth. He was accompanied by his parents. On taking detailed history, it was found that parents had not ever consulted any doctor. Patient was able to ambulate independently with a single stick. He was not having any other complaint as such and was mentally normal. He was having no other physical anomalies in the skull, facies, chest and lumbar region.

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On examination of both the lower limbs, it was seen that pelvis and both femur were normal. The left popliteal fossa was at a lower level than the right. Fusion of the lower limbs was noted at the level of distal one-third of the leg about 8 cm above bilateral ankle joint. Below the level of fusion the right distal leg was straight but the left distal leg and ankle foot complex was deviated laterally with acute angulation. Both feet were maldeveloped with rockerbottom deformity with the absence of great toe on the left foot and few other phalanges in both feet (Figure 1 and 2). On palpation, it was felt that the proximal part of the left tibia was fused with the right tibia. X-rays confirmed our findings of fusion of bilateral tibia with acute angulation of the left tibia with partial absence of metatarsals and phalanges (Figure 3). The development of feet above the level of fusion was adequate but below this level both feet were maldeveloped. A provisional diagnosis of sirenomelia was made and further workup and plan for surgery was made. The parents agreed for workup and left to come prepared for admission but they did not come for admission and also did not respond to repeated calls.

DISCUSSION

A mermaid is a legendary aquatic creature with the upper body of a female human and the tail of a fish. Mermaids are associated with the biological order sirenia comprising dugongs and manatees. Hence this syndrome is named sirenomelia. Sirenomelia is a congenital structural anomaly characterized by abnormal development of the caudal region of the body with varying degrees of fusion of lower limbs [1]. It bears the resemblance of Mermaid of Greek mythology and hence the synonym of Mermaid syndrome [2]. The prevalence of this anomaly is 1:100,000 live births with a male to female ratio of 3:1. About 300 cases are reported in which nine are from India [3].

There are clinically mild and severe varieties. In mild variety, the sirenomelia baby has two limbs fused into one, only to the extent of the skin. The feet may be fully formed and many are only attached at the ankles. All the three main bones of the leg are fully and correctly formed. In this situation, a small surgery can easily correct the deformity whereas the severe variety is very difficult to manage. Externally both limbs are completely joined and appear ill-formed. There is a complete absence of foot structures and out of the three long bones, only two are present in the entire limb. Other internal abnormalities can only be accessed with imaging studies. Distinction was made between these due to the fact that the sirenomelia had a specific pathogenic factor namely arterial steal phenomenon. On the other hand, the caudal regression syndrome was probably due to diabetic embryopathy. An important finding which differentiates these two entities is presence of single umbilical and persistent vitelline artery. Sirenomelia has also been classified into three



Figure 1: Clinical photograph of both lower limbs showing fusion of lower leg with both ill formed feet.



Figure 2: Clinical photograph of both feet with legs showing standing attitude with left popliteal fossa at a lower level than right.



Figure 3: X-ray features of both lower limbs showing fusion of distal part of left tibia with the right with lateral bending of left leg.

types *simpus apus* (no feet, one tibia, one femur), *simpus unipus* (one foot, two femur, two tibia, two fibula), *simpus dipus* (two feet and two fused legs (flipper like)-this is called a mermaid). The first case of sirenómelia was seen in 1542. Duhamel gave the term caudal regression syndrome in 1961 in which he described that sirenómelia was associated with anorectal, genitourinary and vertebral anomalies.

The precise etiology of sirenómelia is not well understood. Many theories have been proposed but none of these is considered conclusive. Hibelink et al. [4] told IV cadmium and lead can produce sirenómelia in the golden hamster. Von Lennep et al. [5] stated teratogenic effects of Vitamin A, Duhammel et al. [6] told manifestation of the caudal regression syndrome. Quan et al. [7] coined the term VATER, Stocker et al. [8] proposed defect in the primitive streak, Stevenson [9] explained vitelline artery steal theory.

Ultrasonography of a fetus with sirenómelia demonstrates the fused femur, decreased distance between two femurs and decreased or absent mobility of the two lower limbs with respect to each other. When infant is clinically examined, there may be only simple fusion of skin of the limbs or there may be fusion of all long bones except fused femur. So imaging studies such as X-rays which show the bony abnormalities and ultrasonography are advised to see for solid organ abnormality. This disorder is universally lethal and hence prenatal diagnosis with imaging studies is very helpful to plan termination of pregnancy [10, 11].

CONCLUSION

In our case, we could not find many of the features as described for this rare condition. Also we could not further investigate to clinch the etiology due to non-cooperation of the patient. This rare abnormality is usually universally fatal but this boy was found to be healthy at fifteen years of age. This case was reported due its rarity.

Author Contributions

Swagat Mahapatra – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Suruchi Ambasta – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor

The corresponding author is the guarantor of submission.

Conflict of Interest

Authors declare no conflict of interest.

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