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Case Report

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SIRENOMELIA: AUTOPSY OF TWO CASES

[1]Daisy Dwivedi and [*2]Bal Chander

[1]Senior Resident, Department of Anatomy, Dr. Rajendra Prasad Government Medical College, Kangra at Tanda. India.
[2]Professor Department of Pathology, Dr. Rajendra Prasad Government Medical College, Kangra at Tanda. India.

***Corresponding Author: Dr. Yemié Aby Alain**

Professor Department of Pathology, Dr. Rajendra Prasad Government Medical College, Kangra at Tanda. India.

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ABSTRACT

Sirenomelia is one of the unusual congenital disorders characterized by varying degree of lower limbs fusion along with visceral anomalies. We are presenting two cases with stress on the lower limb musculature and vascularity. Almost all major thigh muscles could be identified except sartorius which was not well delineated. A single midline artery starting from pubic region and ending around distal ends of tibia and fibula was noted. The artery bifurcated into larger anterior and smaller posterior branches around upper thigh. The anterior branch further bifurcated below knee into larger anterior and smaller posterior branches. Femur along with tibia and fibula were present in pairs. Owing to abnormal vascular features, such cases can be challenging to manage.

KEYWORDS: Sirenomelia; Autopsy; Dissection; Malformations.

INTRODUCTION

Sirenomelia or mermaid syndrome is one of the unusual congenital disorders characterized by varying degree of fused muscoluskeletal structures of lower limbs along with anomalies involving vertebral column, gastrointestinal and genitourinary tract among others.[1] The syndrome is considered to be fatal however there are case reports of live birth some of them even corrected with varying degree of success.[2] The signature malformation involving fusion of lower limbs can range from mild form where all the bones of two lower limbs are discernible on radiological examination to the severest form with a single set of fused bones and rudimentary limb.[3] The frequency of sirenomelia varies from 1.1 to 4.2 per 100,000 births.[4,5,6] Incompatibility with life in most of the cases is because of associated visceral anomalies in the form of renal agenesis and imperforate anus. The surgical correction whenever attempted involves radical and multiple procedures to the correct multiple malformations. We are presenting two such cases both of which had died in utero. X rays could not be done in either of the cases.

MATERIAL AND METHODS

Two cases of Sirenomelia were included for study and after a detailed history including relevant clinical information, autopsy was done in both the cases with particular attention to lower limb dissection.

RESULTS

Case 01

A 29 years old second gravida with uneventful first pregnancy and a normal three years old female child had registered with second pregnancy. A diagnosis of intrauterine death was made on the second visit to the hospital. There was no history of consanguinity, un-prescribed drug intake or radiation exposure. A 24 weeks old dead male fetus was delivered. A complete autopsy was carried out. The fetus weighed 390 grams and the crown rump length was 15 cm. On external examination there was a fused lower limb (Fig 01A) imperforate anus and presence of skin tag in place of external genitalia, In addition there were prominent epicanthic folds and broad flattened nose. There were number of visceral anomalies including blind ending distended colon terminating at superior aspect of distended urinary bladder (Fig 01 B), bilateral multicystic kidneys (Fig 01C) with right hydroureter (Fig 01D). The scrotum was absent along with prostate. The rest of the visceral organs were normal including three umbilical vessels. The lower limbs revealed single vessel coursing along the midline. (Fig 01C inset) Femur, tibia and fibula were present in pairs. (Fig 01 D inset) Fig: 01.

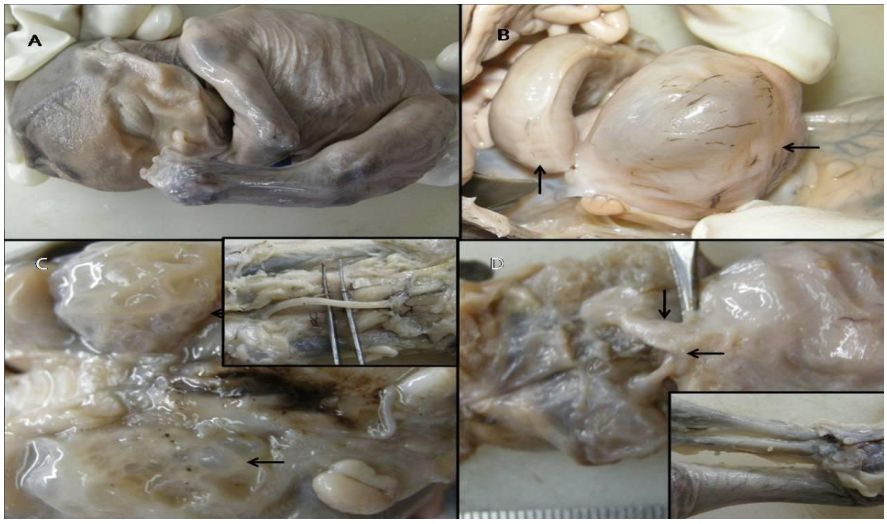

Figure 01: Case 01 gross picture, A; Blind ending distended colon ((vertical arrow) terminating at superior aspect of distended urinary bladder, B; bilateral multicystic kidneys, C; Right hydroureter (vertical arrow), D; Lower limb single vessel coursing along the midline, C inset; Tibia and fibula present in pair, D inset.

Case 02

A 25 years old primigravida registered in the hospital and similar to the case 01 a diagnosis of intrauterine death was made and fetus was sent for autopsy. The pregnancy had been uneventful and there was no adverse history. The fetus weighed 375 grams and crown rump length was 13 cm. The lower fused limb was similar to the case 01 and so was imperforate anus along with blind ending colon blind ending distended colon terminating at superior aspect of distended urinary bladder (Fig 02 A). However apart from multicystic kidneys there were no other malformations. The umbilical cord had two vessels. On dissection of lower limbs all major muscles of thigh including rectus femoris, vastus lateralis, vastus intermedium, vastus medialis, the adductors including adductor magnus and longus, hamstrings including semimembronosis, semi tendinosis and biceps femoris could be identified. All of these muscles were present in pairs except sartorius which was not well delineated. Blind ending distended colon terminating at superior aspect of distended urinary bladder (Fig 02 B) Dissection of lower limb showed an artery similar to case 01 which bifurcated at the area corresponding to pubic region into anterior and posterior main branches. (Fig 02 C) The anterior branch continued along the midline below knee where once again it bifurcated into anterior and posterior branches. The anterior branch below the knee ended in one of the dorsalis pedis arteries. (Fig 02 D) The source of other dorsalis pedis could not be identified. Both the posterior branches, around the pubic region and below knee were smaller and it was difficult to see subsequent branching of the same. Area corresponding to femoral triangle revealed femoral nerves and its branches. (Fig 02 E and F). Fig: 02.

Figure 02: Case 02 gross picture, A; Lower limb musculature, B; Midline artery, bifurcating at the area corresponding to pubic region into anterior and posterior main branches (horizontal arrows), C; One of the dorsalis pedis arteries, D; Area corresponding to femoral triangle with femoral nerves and its branches, E and F.

DISCUSSION

The etiology of sirenomelia is unknown. In humans, sirenomelia may be an autosomal dominant genetic condition and every single case is caused by a new spontaneous mutation. With the limited number of cases in literature, there appears to be some genetic predisposition and twinning is claimed to increase the probability by 100 to 150 times.[6] The frequency appears to be more in female below 20 and above 40 years.[7] According to a few authors, sirenomelia is considered as one of the extreme manifestation of caudal regression syndrome which denotes impaired development of lower half of body including genitourinary and gastrointestinal tract along with lower back and limbs. However others have differentiated sirenomelia from caudal regression syndrome on the grounds that single umbilical artery, lethal renal dysgenesis, oligohydramnios and other related disorders are more common in the former than latter.[7]

Exposure to heavy metals is associated with caudal dysgenesis and sirenomelia in both experimental models and in humans. Maternal diabetes is a causative environmental factor for caudal dysgenesis in 10–15% of affected children. However, this association remains controversial for sirenomelia because only 0.5%–3.7% of reported cases has diabetic mothers.[1,5]

Sirenomelia has been categorized according to the extent of lower limb musculoskeletal fusion. Accordingly Stocker and Heifetz had classified sirenomelia into seven different categories with type I where two complete sets of lower limb bones are discernible to Type VII with single lower limb fused bone and rudimentary limb without differentiation of feet.*[8]* Given that in both the cases presented here femur, tibia and fibula were present in pairs, the cases can be classified as Sympus dipus or symmelia, Type I. In addition to the signature appearance there are host of associated visceral anomalies making it a multi systemic disorder. Commonly observed malformations are lumbosacral and pelvic, malformed or absent external genitalia, renal agenesis, imperforate anus with blind ending colon along with upper body malformations in the form of cardiopulmonary defects and cleft lip or palate.[2,4] It is not surprising therefore that most of the cases result in fetal loss.[2] However there are a few case reports of live births and attempted surgical corrections.[9] Both the cases presented here showed blind ending colon, imperforate anus and multicystic kidneys which might be responsible for intrauterine deaths.

There are different hypothesis regarding etiopathology of sirenomelia. The vascular steal hypothesis is based on the aberrant vasculature pattern of the caudal body present in most individuals with sirenomelia along with single umbilical artery (SUA).[10,11] However it should be noted incidence of that most fetuses with SUA do not have sirenomelia. The incidence of Sirenomelia and SUA is 1 in 200 and 4 per 100,000 respectively.[4,5,6,12] Therefore it is not possible to conclusively prove the causation since not all the cases of sirenomelia have SUA and only a minor fraction of SUA have sirenomelia.

According to the defective blastogenesis hypothesis, sirenomelia is a primary defect of blastogenesis that occurs during the final stages of gastrulation at the tailbud stage, resulting in malformed caudal region and types of sirenomelia is dictated by extent and timing of injury to the said region during embryogenesis.[13,14,15] It is quite possible that defective blastogenesis because of any reason leads to malformations of both the vascular and other tissue resulting in vascular malformations including SUA.

Regardless of the hypothesis which cannot be proven either way in a manner unequivocal, most cases of sirenomelia are not compatible with survival. The visceral malformations seen in the cases presented here including bilateral multicystic kidneys, ureteral and vascular abnormalities along with imperforate anus and blind ending colon clearly indicate that normal visceral functions are not possible and therefore prolonged survival. In principle lower limbs in both of these cases could have been corrected surgically since all the major thigh muscles were identified along with femoral nerves. The correction of vascular component would have been challenging given a single major midline artery.

Keeping in view multiple malformations and poor prognosis in vast majority of cases, early diagnosis is important. A decision to continue pregnancy or otherwise usually requires ethical considerations and entails detailed parental counseling.

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