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Case Report

A CASE OF SIRENOMELIA FOUND DURING STUDY ON FOETUSES

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ABSTRACT

Sirenomelia is a severe malformation of the lower half of the body characterized by fusion of the lower limbs and, commonly associated with renal agenesis, absent external genitalia and other gastrointestinal defects. It has an incidence of one in 60,000 to 70,000 pregnancies. Despite recent progress in pathology, the etiopathogenesis of sirenomelia is still debated, although the assumption that it can have a genetic basis in mice may help in understanding its pathogenesis. Twelve foetuses were used for the study, which were obtained from the department of Obstetrics & Gynaecology of our college. The foetuses were examined for any abnormalities externally later they were dissected to search for any internal abnormalities. One of the foetus showed features of sirenomelia, the foetus was one of the twins of a 22yr old female at 33weeks+3days of gestational age, with history of maternal diabetes, who was admitted for elective caesarean section. The baby died 2 hrs after delivery; the other twin was reported to be normal. The X-ray of the foetus was taken and later dissected. We found abnormalities in the urinary, gastrointestinal and in cardiovascular system.

Key words: Sirenomelia**,** gastrointestinal defects, renal agenesis, external genitalia, cardiovascular

INTRODUCTION

Sirenomelia, the Mermaid Syndrome is a rare and lethal congenital anomaly which is characterized by fusion of the legs and a variable combination of visceral abnormalities, the resultant infant bears a resemblance to the mermaid of ancient Greek mythology[1]. It has an incidence varying between 0.8 to 1 case per 1, 00,000 births[2]. This syndrome has a strong association with maternal diabetes where relative risk is 1:200-250 and up to 22% of foetuses with this anomaly will have diabetic mothers.[3] This anomaly predominantly affects males (sex ratio 2.7:1), and is frequent among one of two monozygotic twins[4].

Its classification as a variant of caudal regression syndrome is still debated, despite the conclusions of Stevenson, the same applies regarding its relationship with narrow pelvis syndrome and VATER (vertebral defect, anal atresia, interauricular communication; interventricular communication, tracheal and esophageal atresia, and renal or radial agenesis) syndrome. It is usually incompatible with life, yet there are a number of reported cases of surviving infants with this condition in the English literature[5]. Its ethiopathogenesis remains controversial.

MATERIALS AND METHODS

Twelve foetuses were used for the study which were obtained from the department of Obstetrics & Gynaecology of our college. The foetuses were examined for any abnormalities externally later they were dissected to search for any internal abnormalities.

One of the male foetus showed features of sirenomelia, the foetus was one of the twins of a 22yr old female at 34weeks of gestational age, with history of maternal diabetes, who was admitted for elective caesarean section. The baby died 2 hrs after delivery; the other twin was reported to be normal. The X-ray of the foetus was taken and later dissected. We found abnormalities in the urinary, gastrointestinal and in cardiovascular system.

OBSERVATION

Externally there was fused lower segment of the body below the pelvis into a single lower limb, with two feet fused posteriorly giving a single flipper-like foot with nine toes spread out in a fan-like pattern, the foot was oriented anteriorly relative to the trunk there was fusion of both the lower limbs (fig1).

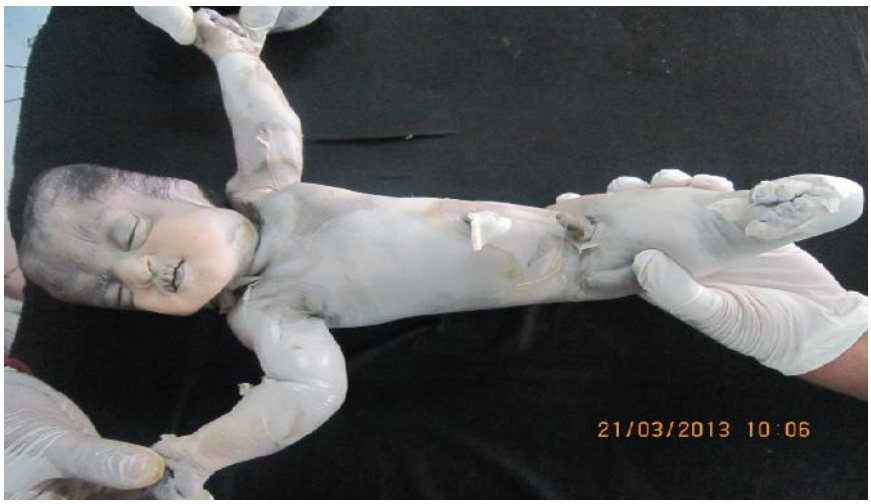

Fig: 1

2. A single umbilical artery was seen (fig2).

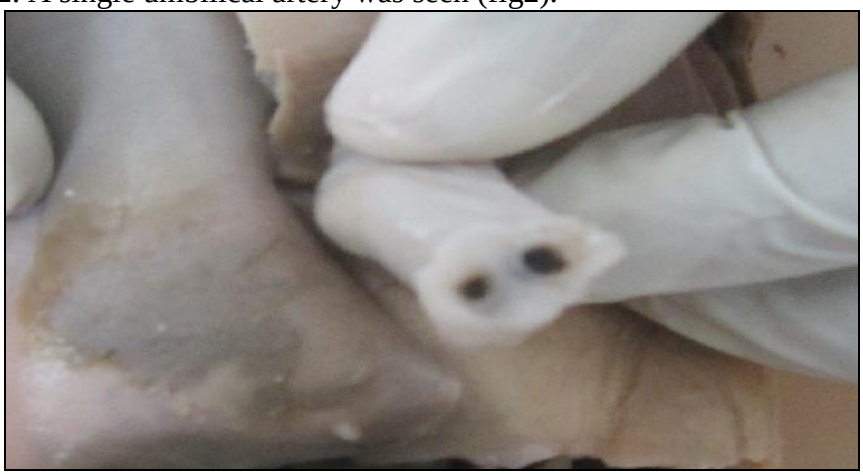

Fig 2

3. Imperforate anus was found, external genitalia were found to be normal, and there was absence of testes on palpation in the scrotum (fig3).

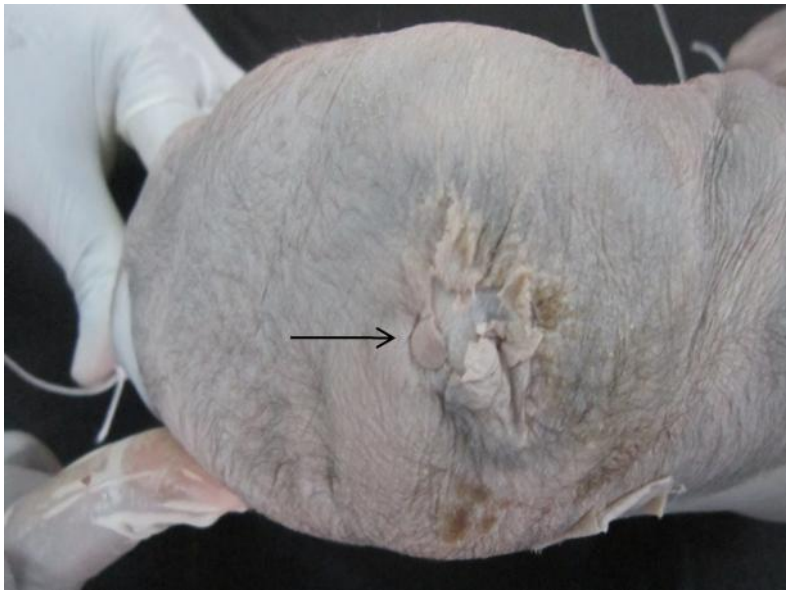

Fig:3

On dissection internally we found (i) Absence of kidneys and ureters bilaterally (ii)Absence of urinary bladder (iii) Adrenal gland was also absent bilaterally (iv) In the GIT there was found a blind ending colon (fig 4)

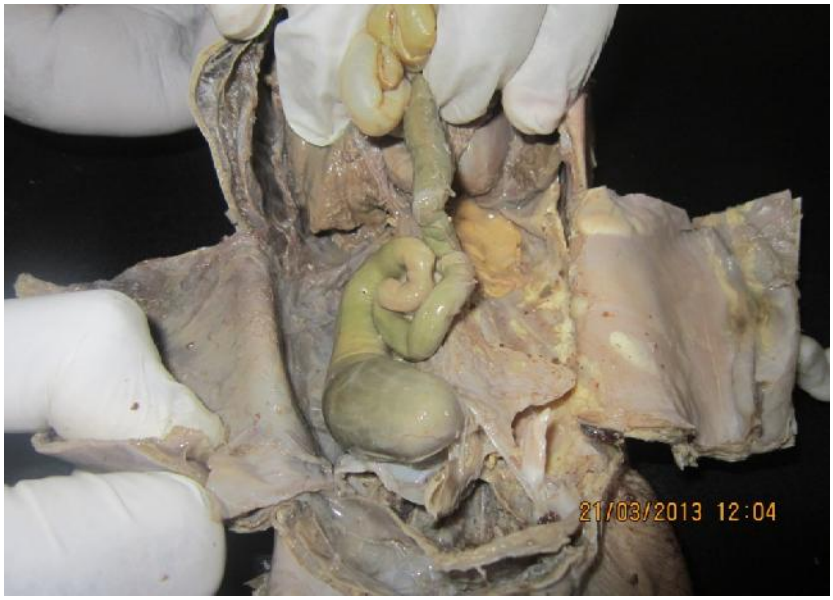

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Fig: 4

v) The abdominal aorta ended as a single left common iliac artery. (vi) In the thorax we found the left vertebral artery was arising from the arch of aorta (fig 5) and there was a patent foramen ovale (fig 6)

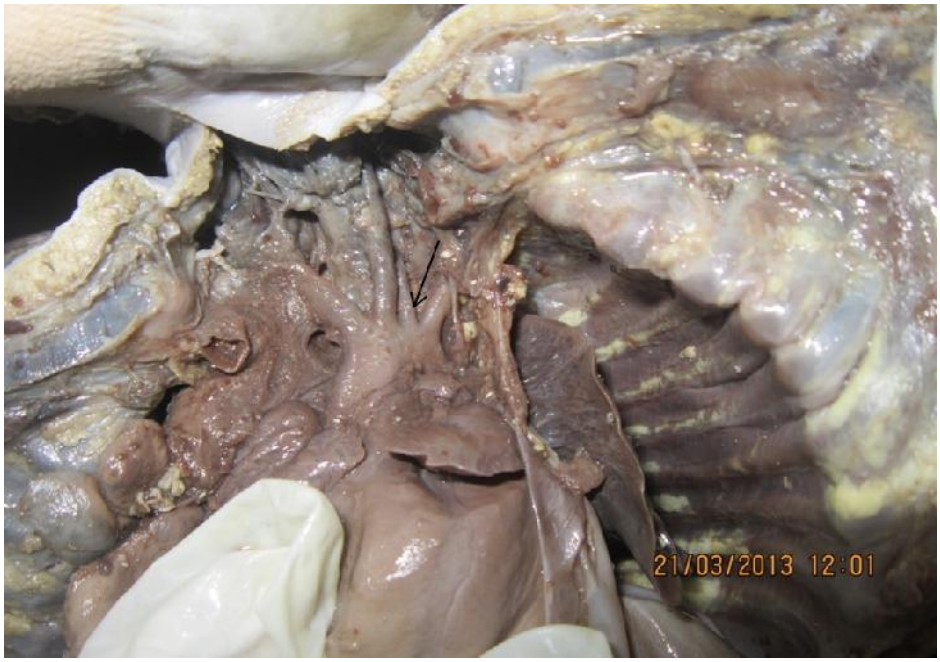

Fig: 5

Fig: 6

X ray showed presence of fibulae medially placed, no other skeletal anomalies was seen.(fig 7)

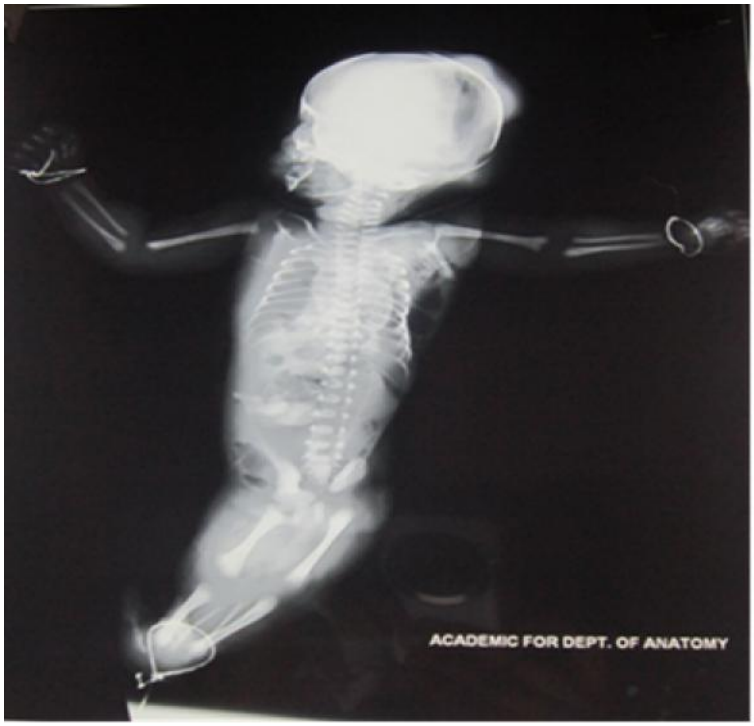

Fig:7

DISCUSSION

Sirenomelia was initially described by Rocheus in 1542 and Palfyn in 1553, till date approximately 300 cases have been reported in the literature[2,3]. The diagnosis, which is obvious at birth, is currently performed by prenatal ultrasonography, oligohydramnios in the second trimester is the alerting sign[6], clues also include renal agenesis and a fibula positioned between the tibiae

Sirenomelia has been classified into three types according to the number of lower limb bones present:

1. Sirenomelia apus: No feet only one tibia and one femur.
2. Sirenomelia unipus : One foot, two femurs two tibia, and two fibula.
3. Sirenomelia dipus: Two feet and two fused legs giving the appearance of a flipper [2].

From developmental point of view, the sequence of events leading to sirenomelia may be the results from an 'embryological insult' involving the caudal mesoderm occurring between days 28–32 of foetal life. By this gestational age, the cloaca is already formed, the kidneys are found in the pelvis while the gonads are intra-abdominal. Hence any developmental abnormalities of the caudal extremity during this period would affect equally the kidneys, the bladder, the terminal bowels, the pelvic bones as well as the genitalia (excluding the gonads which are intra-abdominal) [4]

Stevenson *et al* proposed the vascular steal theory to explain the development of abnormalities on the caudal extremity. This theory suggests that there is shunting of blood via an abnormal abdominal artery arising from high up in the aorta towards the placenta. Consequently there is hypoplasia of the vasculature distal to the artery leading to nutritional deficiency of the caudal half of the body. Hence there may be complete or incomplete

agenesis of the caudal structures described above, except the gonads which are intra-abdominal[7]. The single umbilical artery in our patient favors' this theory.

A third theory regards sirenomelia as part of the caudal regression syndrome (CRS). This is a rare congenital defect characterized by a broad spectrum of lumbosacral agenesis which was described by Duhamel to include genitourinary and vertebral anomalies[8].

Maternal diabetes is the only maternal disease known to be associated with sirenomelia (2% of cases), Welch et al suggested an association between maternal diabetes and a genetic predisposition[9].

Teratogens like retinoic acid, cadmium and cyclophosphamide have been implicated in the genesis of sirenomelia in mice and hamsters. However, no case of sirenomelia in humans has been observed after incidental maternal exposure to these products[5]. Sirenomelia occurs in mice lacking Cyp26a1, an enzyme that degrades retinoic acid (RA), and in mice that develop with reduced bone morphogenetic protein (Bmp) signaling in the caudal embryonic region. The phenotypes of these mutant mice suggest that sirenomelia in humans is associated with an excess of RA signaling and a deficit in Bmp signaling in the caudal body[10].

Recent reports indicate that about 50% of these infants are born alive after 8–9 months gestation, however most of them die within 5 days of life, survival depends on the associated visceral anomalies, especially renal function, rather than the sirenomelia itself[4]. Initial treatment of these newborns includes supportive care and diverting colostomy, later management of these infants includes a multidisciplinary surgical approach involving various specialties [2].

CONCLUSION

Sirenomelia, being a rare anomaly emphasis should be placed on prenatal diagnosis to minimize the trauma related to the termination of pregnancy at advanced gestation and ensure an optimal management that would consequently be less demanding both from a psychological point of view and a health cost point of view. The information lately gathered from animal models, suggests a complex multigenetic basis, hence the future should include research to assess the role of these genetic factors in the generation of this malformation.

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