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Case Report

SIRENOMELIA: MERMAID SYNDROME

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ABSTRACT

A neonate with rare congenital anomalies was born at 25 weeks of gestation and died within 17 minutes of birth. On examination of the baby, it was found that the lower limbs were malrotated and fused all along the length with six toes. External genitalia, urogenital and anal orifices were absent. At autopsy, a single umbilical artery was found arising from the abdominal aorta. Both the kidneys were polycystic and were situated in the iliac fossae. Distal portion of the large gut beyond caecum was absent and rectum was atretic. No reproductive organ was found. On the basis of the findings, the case was diagnosed as sirenomelia (mermaid syndrome).

KEYWORDS: Sirenomelia, polycystic kidneys, single umbilical artery.

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INTRODUCTION

Sirenomelia (mermaid syndrome) is an extremely rare congenital disease leading to formation of internal and external defects in the process of development of the human body [1]. Originally described by Rocheus in 1542 and Palfyn in 1543 [2,3], this syndrome is characterized by fusion of lower limbs which is also associated with anomalies like flexion, external rotation, symelia and atrophy [2,3]. Moreover, in this syndrome, there are some lethal defects such as agenesis or any other anomaly of one or both kidneys as well as other gastrointestinal defects [1]. Another pathognomonic finding is the presence of a single umbilical artery which is the chief distinguishing finding from Caudal Regression Syndrome [4]. The incidence of this syndrome varies between 0.8 and 1 per 100,000 births [5].

CASE REPORT

A 24 year old mother,$P_{0=1,}$ presented with pain abdomen at 25 weeks of gestation. She gave a past history of abortion and irregular intake of oral contraceptives. She was non-diabetic and non-hypertensive. She had a family history of diabetes, but no incidence of any congenital anomaly or stillbirth. Antenatal ultrasound scan was done prior to the admission of the mother in the hospital. The scan reported the presence of a single live breech baby with severe oligohydramnios. Polycystic changes were noted in both the kidneys of the fetus, No remark was there about any other abnormality. Placenta was fundal with grade II maturity. The mother underwent assisted breech delivery in a State Government Hospital. She delivered a baby weighing 1.2 kgs. with fused lower limb.

The baby did not cry after resuscitation and died within 17 minutes of birth.

At birth: The baby presented Potter's facies characterized by prominent infraorbital folds. Lower limb was found to be single, malrotated, fused all along the length with six toes (Fig.1). Flexion at the hip joint was also found. There were no external genitalia, urogenital or anal orifices. The upper limbs showed syndactyly and brachydactyly.

X-ray (post mortem) of the baby: It revealed presence of only femur and tibia and absence of fibula on both sides (Fig.5).

At autopsy: A single umbilical artery was found (Fig.2) arising from the abdominal aorta. The aorta was absent below the origin of the umbilical artery. Kidneys were present in the iliac fossae. The kidneys were polycystic macroscopically and were supplied by branches from the abdominal aorta (Fig.3). Bladder was rudimentary with two ureters opening as usual into it. The small intestine was normal. The caecum was dilated. As there was no ascending colon, the caecum ended in a blind loop on both sides (Fig.4). The distal portion of the large gut beyond the caecum was absent upto the rectum which was found to be atretic. An insignificant small dimple was seen at the region of the anal orifice. No gonad was present. No significant reproductive organ could be seen either. Karyotyping could not be done.

Fig. 1: Stillborn diagnosed as sirenmelia (mermaid syndrome).

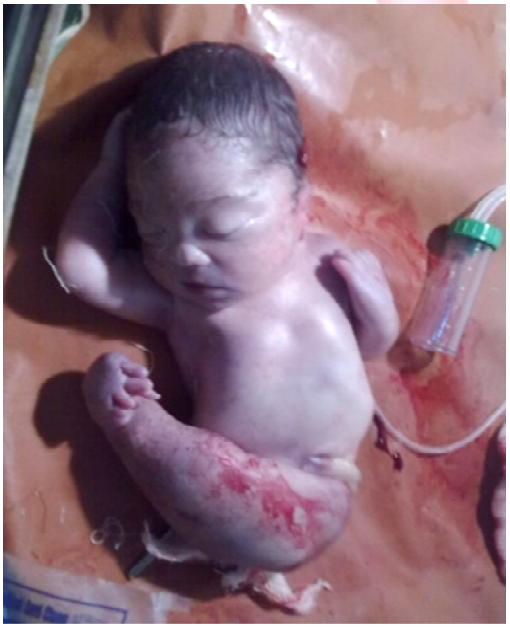

Fig. 2: Polycystic Kidneys (thick arrows) and single umbilical artery (thin arrow) at autopsy.

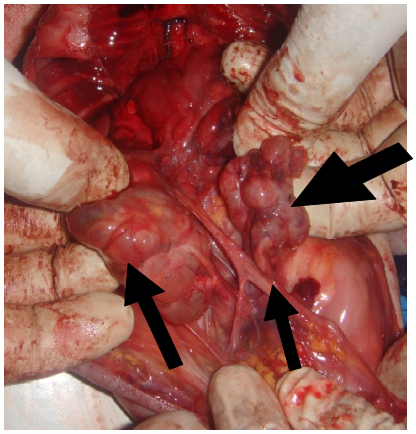

Fig. 3: Polycystic kidneys (thick arrows) supplied by branches (thin arrows) from the abdominal aorta.

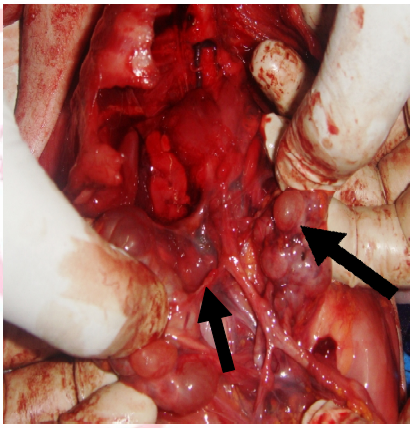

Fig. 4: Normal small gut (thin arrow) and dilated caecum (thick arrow) at autopsy.

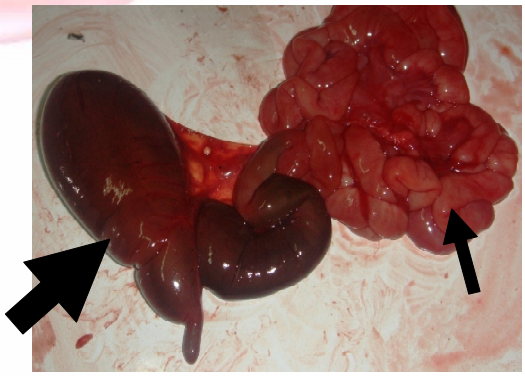

Fig. 5: Radiograph showing the presence of only femur and tibia and absence of fibula on both sides.

DISCUSSION AND CONCLUSION

Being a rarest of the rare kind of anomaly, sirenomelia has approximately 300 reported cases in the literature till today [5]. Most of the reported cases here have been diagnosed after delivery as no definitive diagnosis was possible antenatally. Etiology of this syndrome can be multifaceted of which one of them is teratogenicity.[6]Animal study confirmed the association with retinoic acid [7], cocaine [8] or irradiation [9], though convincing data regarding this fact are few. Intracytoplasmic sperm injection is also considered as a causal factor [10].

In the present case, the affected baby was diagnosed as a case of sirenomelia (mermaid syndrome) based on physical features, ultrasonography, radiology and autopsy findings. There was a maternal history of tobacco chewing which could be a probable risk factor in the birth of a mermaid baby. Moreover; the occurrence of this type of anomaly strongly suggests ritual antenatal screening of the mother at regular basis and also advice regarding the drawbacks and use of oral contraceptives judiciously.

Conflicts of Interests: **None**

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