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Sirenomelia-A rare fetal anomaly: Case report

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ABSTRACT

Sirenomelia, the Mermaid Syndrome is a rare and lethal congenital anomaly with an incidence of 1 in 60,000 to 70,000 pregnancies. Sirenomelia is characterized by complete fusion of the lower limbs, commonly associated with renal agenesis, absent external genitalia and other gastrointestinal defects. Another pathognomonic finding is the presence of single umbilical artery (persistent vitelline artery) which is the chief distinguishing anatomic finding from Caudal Regression Syndrome. We report a case of rare fetal anomaly which was identified to have characteristic features of Sirenomelia at the time of delivery.

Keywords: Sirenomelia, Mermaid syndrome

INTRODUCTION

Sirenomelia Sequence [Mermaid] is a congenital structural anomaly characterized by an abnormal development of the caudal region of the body with different degrees of fusion of the lower extremities and bears resemblance to the mermaid of Greek mythology. This deformity is also known as Symmelia, Symposia, Sympus, Uromelia and Monopodia. Sirenomelia has a prevalence of two to three cases per one lakh births with male: female ratio of 3:1. About 300 cases have been reported in the world literature so far of which eight have been reported in India.[1]

Sirenomelia has been classified as a)Simpus apus: No feet , one tibia, one femur b) Simpus unipus : One feet ,two femur ,two tibia , two fibula c) Simpus Dipus: Two feet and two fused legs (flipper like) - this is called a mermaid.[1]

The precise etiology of Sirenomelia is not well understood. Many theories have been proposed but none of these is considered conclusive .[1]

Prenatal diagnosis of Sirenomelia is possible by demonstrating the fused femur, decreased distance between two femur and decreased or absent mobility of the two lower limbs with respect to each other. On physical examination of the infant, the defect varies from simple cutaneous fusion of the limbs to absence of all long bones except fused femur. Radiograph of the infant shows the exact bony anomaly while USG abdomen can demonstrate the solid viscera. Thus, it is a universally lethal entity and prenatal diagnosis on USG is desired so that termination of pregnancy can be offered at the earliest.[1] We report a rare such case of sirenomelia diagnosed at birth.

A 25 year old gravida 5 para 3,live 2 presents with labor pains .No history of consanguinity, with history of one abortion in first trimester, one still birth with polyhydramnios ,present pregnancy had no antenatal scans done ,all

the trimesters were uneventful. Emergency Caeserean section was done due to premature rupture of membranes and an anomalous baby delivered , which cried at birth but expired later.

Ultrasound scan done at that time revealed single umblical artery with?? Renal agenesis.Post mortem fetal X-Ray revealed single femur bone. (Fig 1 &2).Later, the foetus was sent for autopsy.

Gross examination revealed a 2.5 kg weighing baby with under developed caudal stump with illformed labial folds(Figure 3& 4). Umblical cord measuring 5.5 cm in length

MATERIALS AND METHODS

After autopsy, the fetus was fixed in 10%formalin for processing. After gross analysis representative sections were given for tissue processing .Sections were processed routinely with paraffin embedding and stained with haematoxylin and eosin.

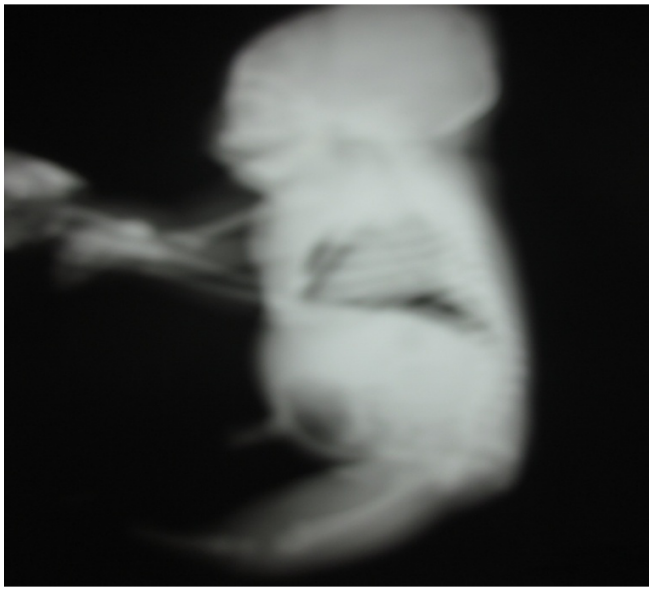

Figure 1: Fetal X-Ray – post partum reveals single femur bone

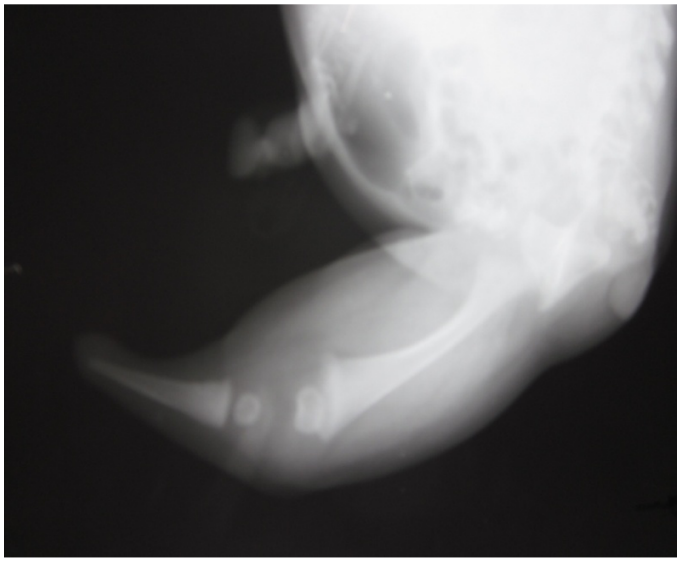

Figure 2: : Fetal X-Ray – post partum reveals single femur with absent fibula, rudimentary sacrum and iliac bones

Figure 3: Fused lower limbs with caudal stump

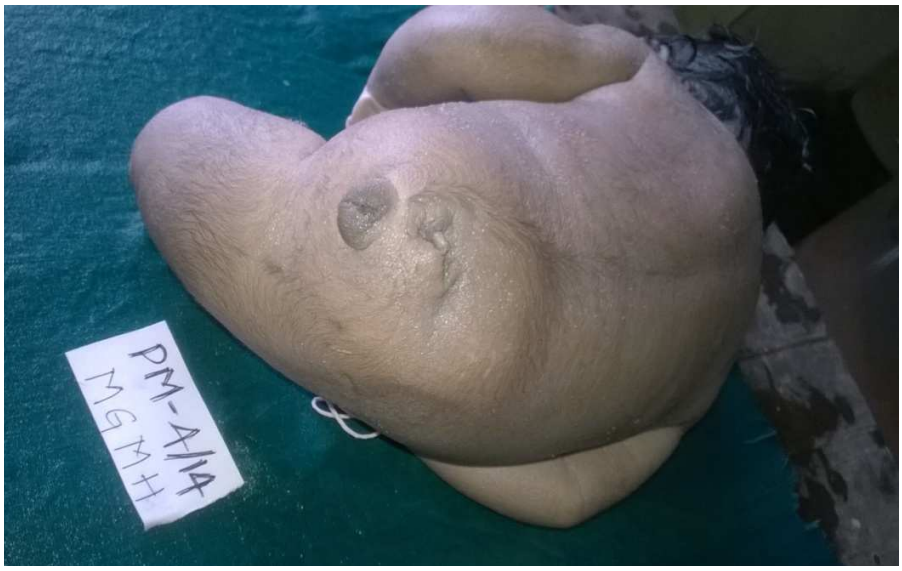

Figure 4:Ventral aspect showing caudal stump with ill formed labial folds and anal atresia

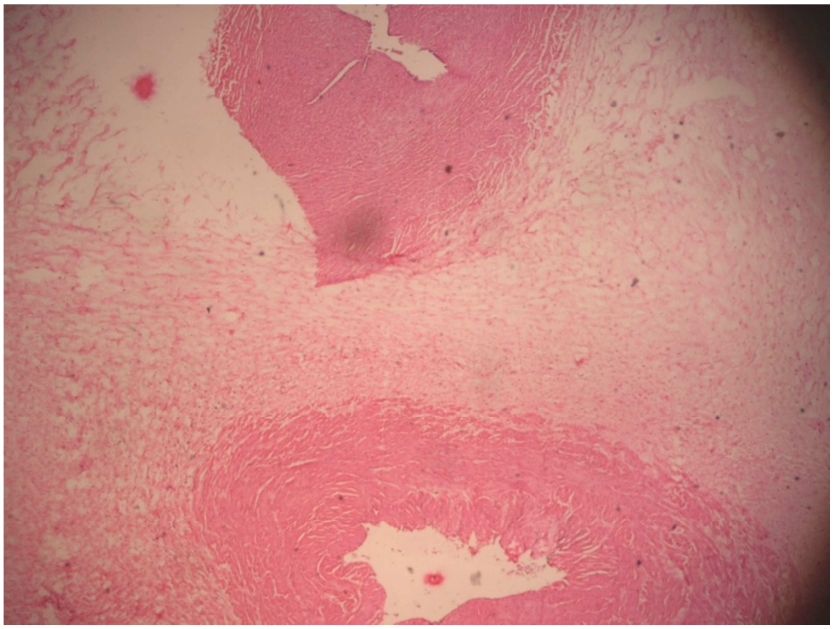

Figure 5: Histopathological examination of the sections of the umbilical cord show single umbilical artery and vein

The diagnosis of Sirenomelia was made based on the X-Ray findings of single femur with absent fibula, rudimentary sacrum and iliac bones , autopsy gross findings of fused lower limbs replaced by caudal stump with ill formed labial folds and anal atresia. The histopathological examination of umbilical cord revealed a single artery and vein.

RESULTS AND DISCUSSION

Sirenomelia is a rare anomaly of caudal region of the body presented with fusion of lower limbs in which Genito-urinary, Gastrointestinal, Cardiovascular and Neural tube anomalies are found in most cases.[2] Etiology of sirenomelia is not well understood[2]. It is a rare syndrome (0.8 to 4.2/100,000 births) which continues to cause many controversies concerning its etiopathogenesis. .Five pathogenetic theories of sirenomelia are described like an embryological insult ,vascular steal theory,as part of the caudal regresion syndrome, as part of the VACTERL syndrome (vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies, and limb abnormalites) and external forces acting on the caudal extremity.

From an embryological point of view, the sirenomelia sequence comes from an ‘caudal mesoderm damage taking place betwen days 28–32 of fetal life (first theory). Therefore there are renal agenesis, lack of genital organs, imperforated anus, vertebral dysgenesis, and lower limb atrophy.[2] The diagnosis which is obvious at birth, is currently performed by prenatal ultrasonography

CONCLUSION

Our experience and the available literature support the fact that careful pathological examination of foetus can confirm clinical diagnosis and to explain the cause of intrauterine fetal demise.

It helps to identify unexpected anomalies that may provide further clues to a diagnostic syndrome and also assist in family planning, genetic counselling for future pregnancies.

Thus, future attempts can be made to develop a simple and reliable method for obtaining fetal DNA from maternal blood in establishing prenatal diagnosis. [3]

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