

Case Report

Sirenomelia: Case Report and Discussion of its Prenatal Diagnosis

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ABSTRACT

Sirenomelia is a rare polymalformative, lethal congenital anomaly, characterized by fusion of both lower limbs, bilateral renal agenesis, absent external genitalia, aberrant fetal vasculature, and severe oligohydramnios. We hereby report a case of Sirenomelia diagnosed postnatally in an unbooked pregnancy presenting at the thirty-fifth gestational week.

KEY WORDS: Diabetes mellitus, dysmorphic lower limb, prenatal diagnosis, sirenomelia

INTRODUCTION

A Sirenomelia sequence is a rare polymalformative syndrome, where the most noticeable (but inconstant) aspect is the fusion (more or less complete) of the lower limb.^[1] With a male predilection and sex ratio of 2.7:1, the anomaly is more common in monozygotic twins.^[2]

As it is a lethal abnormality,^[1] with recurrence risk of 3–5% in subsequent pregnancies, the Obstetricians and Radiologists should be aware of the methods to use for its prenatal diagnosis, and hence, its timely management.

The present report is a case of Sirenomelia in an unbooked antenatal woman presenting with preterm labor at 35 weeks. Diagnosis was made postnatally based on the morphology and radiographic features. Antenatal diagnosis was missed due to severe oligohydramnios. We hereby discuss the embryological theories and methods of prenatal diagnosis of this lethal anomaly.

CASE REPORT

A 25-year-old primigravida, presented as an unbooked pregnancy at 35 weeks, with threatened preterm labor. She took iron and folic acid supplementation along with

Tet-Vac immunization from a local dispensary, but did not get any routine investigation done. There was negative history of any pathological antecedent factor (diabetes or teratogen exposure) or consanguinity. Her vitals were stable. The abdominal examination showed a 30 weeks gravid uterus with single live fetus and mild contractions. Per vaginal examination showed that the patient was in the first stage of labor. An ultrasound examination done before starting tocolysis showed a single live fetus with biometric parameters corresponding to 34 weeks with severe oligohydramnios and absent bladder and kidneys. The absence of amniotic fluid obscured a precise morphological study. In view of a congenitally anomalous fetus, tocolysis was not given and labor was allowed to progress. She delivered, vaginally, a polymalformed fetus, with a 2-kg birth weight, and poor APGAR score, which succumbed after 30 minutes of birth from respiratory distress (probably owing to pulmonary hypoplasia). The placenta was normal with eccentric attachment and a two-vessel cord.

The pathological examination of the neonate showed

1. Morphological abnormalities [Figure 1]
 - Undetermined sex
 - Single lower limb
 - Potter facies
 - Imperforate anus
 - Absent urinary orifice.
2. Radiograph of the skeleton [Figure 2] revealed
 - Partial sacrococcygeal agenesis
 - Single lower limb with normal appearing femur

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Figure 1: Shows the morphological appearance of the fetus with sirenomelia

- Absent fibula
- Short tibia showing distal shaft tapering
- Absent ankle and foot bones (tarsals, metatarsals, and phalanges)

Internal abnormalities could not be evaluated as the parents did not consent for neonatal autopsy. The fetal karyotype was 46XY. The patient's fasting blood sugar post delivery and 75 gram Glucose Tolerance Test done after six weeks were normal.

DISCUSSION

There are methods of diagnosing or at least suspecting the entity. If we go by maternal history, the only maternal disease known to be associated with Sirenomelia is Diabetes Mellitus (2% of diabetic cases). This is in contrast to a Caudal Regression Syndrome (CRS), which is seen in 22% of diabetic cases.^[3] According to the classification by Stanton *et al.*, Sirenomelia is the most severe form in the spectrum, constituted by CRS (also called Caudal Dysplasia Sequence).^[4]

The etiology is still debatable.^[3] Welch *et al.* suggested an associated genetic predisposition.^[5] However, teratogen exposure (cocaine, organic fat solvents, and appetite suppressors like diethylpropion, lead, cadmium, retinoic acid, and radiation) during the gastrulation stage (i.e. during the third gestational week, interfering notochord development leads to abnormal development of the caudal structures) has also been implicated in some cases of CRS in humans.^[1] On account of notochordal maldevelopment, Neural Tube Defects are frequently associated.^[1] According to such a theory, Sirenomelia forms two extremes of a single malformative spectrum.^[2] Moreover, anencephaly and alobar holoprosencephaly have also been associated, explained by multisegmental failure in axial mesodermal patterning. As per

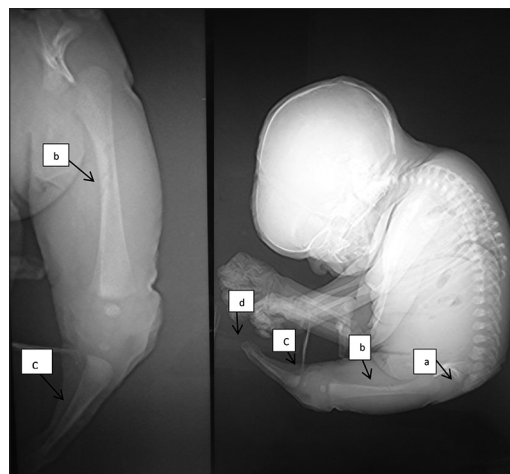


Figure 2: Shows the antero-posterior view of the lower limb and lateral view of the fetal skeleton radiograph showing (a) partial sacrococcygeal agenesis (b) single normal femur (c) short tibia with distal shaft tapering, absent fibula (d) absent tarsals, metatarsals, and phalanges

the Vascular Steal theory by Stevenson, initially known as the nutritional deficit theory (by Kampmeier in 1927), vascular redistribution in the fetus leads to vascular (nutritional) deficit to the embryo's caudal extremity, leading to non/maldevelopment of the caudal extremities. Our observation of a single umbilical artery and renal and bladder agenesis might support this theory. However this theory fails to explain the frequent association like cranial, cardiac, and esophageal defects, and the relationship with the VACTERL complex (Vertebral anomalies, Anal atresia, Cardiac defects, Tracheoesophageal fistula and/or Esophageal atresia, Renal and Radial anomalies, and Limb defects).^[6,7]

Stocker and Heifetz (1987), classified Sirenomelia into seven variants, based on the presence of skeletal elements in the lower limb (LL). Although in Type I (mildest form), all the bones in the two fused LL are present and fusion affects only the superficial tissues, Type VII is the most severe, where only a single femur is present without indication of legs or feet. Type II, III, and IV have a single fibula, absent fibula, and fused fibulae with partially fused femurs, respectively. Type V and VI have partially fused femurs and a single tibia and femur, respectively.

Prognosis-wise, the condition is fatal. Fifty percent of the children are born alive, but mostly succumb to death in the first neonatal week.^[1] Sirenomelia is incompatible with life owing to its association with severe anomalies, for example, bilateral renal agenesis, however, exceptional cases of neonates surviving with minor renal abnormalities or even normal kidneys have been reported by McCoy *et al.* and Clarke *et al.*^[7,8] Sirenomelia dipus, also called as mermaid syndrome, has the most favorable outcome.^[9] Such infants' management needs a multidisciplinary surgical approach, involving various specialties.^[10]

It is thus important for obstetricians and radiologists to have an open eye for this entity in the antenatal period, so that the necessary early intervention can be taken.

The prenatal diagnosis by a reliable sonographic evaluation of the lower extremity in the second and third trimesters is not possible, owing to severe oligohydramnios or anhydramnios, due to bilateral renal agenesis, however, the first trimester or early second trimester scans, due to normal amniotic fluid volume, which is unrelated to fetal urine production, allow detection of abnormalities, namely, anorectal atresia, lumbosacral agenesis, and the like.^[1] In some cases, bilateral renal agenesis is the only antenatal sonographic finding and complete diagnosis is only made after birth.^[1] In our case also, an antenatal diagnosis was missed due to lack of early scans and the severe oligohydramnios detected in the third trimester scan.

Doppler flow imaging can detect a two vessel cord, which is a frequent association. A radiologist should actively look for the sonographic features of Sirenomelia (oligohydramnios, single lower limb, absent bladder, undetermined external genitalia, anorectal atresia, lumbosacral agenesis), in the early anomaly scan and early second trimester scans of all fetuses with bilateral renal agenesis, malformed lower limbs, and a single umbilical artery, which can allow pregnancy termination at an early stage.^[1] Moreover, authors recommend that every sonologist, after taking the femur length, must look for the presence of another femur, during the routine antenatal scans. In our case the condition remained undiagnosed completely till delivery, it being an unsupervised pregnancy. Genetic counseling and an early anomaly scan should be a routine in a pregnant female with related history, as the recurrence risk is 3-5%.^[1] Screening for Sirenomelia in diabetic women and screening of diabetes in pregnant woman with a Sirenomelic fetus is justifiable.^[1]

CONCLUSION

This is a rare entity with controversial etiopathogenesis. Emphasis must be laid upon the prenatal diagnosis, so as to allow optimal early intervention, with minimal psychological and physical discomfort to the patient.

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