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Case Report

Pathology Section

Sirenomelia Apus – A Case Report with Review of Literature

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ABSTRACT

Sirenomelia is a lethal fetal malformation where the lower limbs are fused giving the appearance of woman body and fish tail. It is associated with gastrointestinal and urogenital malformations and is incompatible with postnatal life. Early diagnosis during antenatal period helps in less traumatic termination of pregnancy. Recurrence is unknown / not increased.

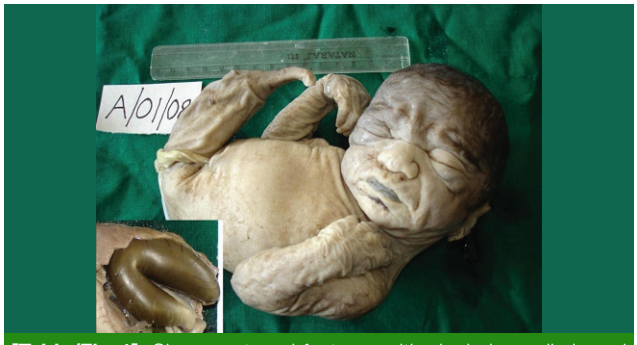
In present case, 19 years female presented with 28 weeks of amenorrhea and antepartum hemorrhage. Ultrasound revealed placenta previa, abruptio placentae and intrauterine death of fetus. Medical termination of pregnancy was done. X-ray of fetus, external examination and internal examination showed features of Sirenomelia – Apus type.

Keywords: Congenital malformation, Fetal anomaly, Mermaid syndrome, Siren.

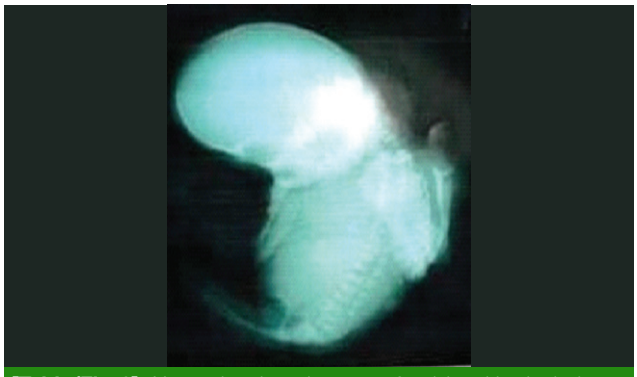
CASE REPORT

A case of 19-years-old female was reported in the Department of Pathology ,with gravida 3, para 2, living 1, death 1, presented with 28 weeks of amenorrhea and antepartum hemorrhage for one day. Family history was not significant. Obstetric history revealed, married life of four years. First pregnancy was preterm vaginal delivery and living female baby. Second pregnancy was intrauterine death of fetus. In present pregnancy, quickening was felt in fifth month. Following per-abdominal examination, a provisional clinical diagnosis of intrauterine death was made. Ultrasound examination revealed abruptio placentae with placenta previa. Medical termination of pregnancy was done and the fetus at birth showed congenital malformation. Consent for autopsy was obtained from the couple.

External examination showed–weight of the baby was 950 grams, significant findings were–fused lower limb without foot measuring 10 cms in length, anal opening and external genitals were not present [Table/Fig-1]. Umbilicus showed single artery. X-ray of fetus showed absence of pelvis with sacral and coccyx bones fused forming a single long bone [Table/Fig-2]. Internal examination revealed; large intestine ending as blind loop with absence of rectum / anal canal and no anal opening. There was bilateral renal agenesis with absence of bladder. All other internal organs were within normal limits with respects to position, gross and microscopy for the age. Final diagnosis of Sirenomelia, Apus-type was made.



[Table/Fig-1]: Shows external features with single lower limb and absence of foot. Inset shows blind loop of large intestine.



[Table/Fig-2]: X-ray showing absence of pelvis with single lower limb

DISCUSSION

Sirenomelia is a rare lethal fetal malformation. It means women body with fish tail. It is a condition where there is a fusion of lower extremities associated with malformation of gastrointestinal and urogenital malformations. It is due to fusion of two posterior appendages giving rise to a single conical mass resembling the body of fish in its posterior half [1-4]. The synonyms are Mermaid syndrome, Siren, Sympodia,

Sympus, Symmelia (fusion of lower extremities), Monopodia and Uromelia [1-3,5,6]. The fetus resembles mythical Siren having amphibian like eyes and ears are floppy and low [7,8]. Duhamel in 1961 first described it as caudal regression / caudal dysplasia [1].

The reported incidence is one in 60,000 to 100,000 live births/ 0.01 – 0.16 per 10,000 live births with male to female ratio of 2.7-3:1. Approximately 300 cases are reported in world literature of which 13 cases are from India. It is 100-150 folds common in one of two monozygotic twins over singleton and dizygotic twins. The incidence is 1 in 100,000 pregnancies [2-5,7,9]. However, in the present case it was single intrauterine pregnancy.

The exact etiology is unknown and may be multi-factorial involving environmental and genetic factors. Environmental factors like physical, chemical, hormonal, teratogens, infections, metabolic and nutritional deficiencies especially vitamin A [2]. The only proved maternal disease associated with Sirenomelai is diabetes mellitus, the reported relative risk is 1:200-250 constituting 2-22% of cases [4,7,10]. Genetic/ chromosomal anomaly is not known and hence it is not hereditary [6,7]. However, Lynch et al., has suggested that 7q36 genes has a role in differentiation of midline mesoderm at both ends of the developing notochord [7]. In a study genetic analysis has shown sporadic mutations [2].

Three theories have been put forth to explain the pathogenesis. The first is the primary failure in developmental of caudal somites due to defect in primitive streak / notochord during third week of embryonic life, derangement in gastrulation that interrupts migration and differentiation of mesoderm resulting in array of defects. The severity of malformation depends on the number of somites that fail to be laid down at caudal aspect of the embryo and specific region / regions of the primitive streak that are disturbed. The kidney defects depend on the mesonephric ducts reaching the uretric buds and metanephric blastema. The notochord acts as neural inducer, neural tube defects are associated with Sirenomelia. This theory explains Sirenomelia and caudal regression syndrome as two ends of the single malformation spectrum, where Sirenomelia is the severe type with renal agenesis and the other end is CRS with very rare / variable disease of renal agenesis [1,2,4,6,9].

The second theory is “vascular steel theory” causing incomplete development of the caudal region of the fetus. Normally blood flow back to placenta through the paired umbilical artery arising from the iliac artery. Where as in Sirenomelia blood returns to the placenta through single large vessel, abnormal persistence of vitelline artery, arising from aorta just below the diaphragm associated with hypoplasia of aortic collaterals downwards especially the renal and inferior mesenteric arteries before it bifurcates into iliac arteries. The defect results in diversion of blood flow from caudal structures of embryo to the placenta with less nutrition and oxygen for development of caudal structures i.e., associated with decreased perfusion, ischemic changes and defective / under development / maldevelopment of lower somites. The defects in blood vessel are variable giving rise to variable spectrum of abnormalities of the structures supplied by distal aorta. Hence no two cases of Sirenomelia are identical. This theory explains urogenital and gastrointestinal defects, however fails to explain cranial, cardiac and esophageal defects. This theory states that vascular anomaly does not occur in CRS and hence Sirenomelia and CRS are two different entities [2,3,9,10].

The third theory is “pressure theory”, which explains that renal agenesis results in oligohydramnios which in turn give rise to caudal pressure effects and abnormalities in caudal region [1,2].

Various defects reported are Potter’s facies, single artery / two vessels in umbilical cord, imperforate anus due to incomplete migration of mesodermal septa along with anorectal atresia, agenesis of urinary bladder due of abnormality of the allantois, bilateral renal agenesis, ambiguious genitalia, agenesis of vertebrae / lumbosacral bone due to failure / malformation of sclerotomes along with varying degree of lower limb fusion and thoracolumbar spinal anomalies, cardiac anomalies, anterior abdominal wall defects and oligohydramnios [1-4]. A case of esophageal atresia is reported [11]. Less than 10% has central nervous system anomalies [7]. In present case the fetus presented with single umbilical artery, no anal opening, anorectal atresia, agenesis of urinary bladder and both kidneys, no external genitalia, absence of pelvis with sacral bone and coccyx fused to form a single long bone without foot.

The abnormality of lower limb can be a simple fusion of lower limb by soft tissues to presence of single, rudimentary limb. The fused legs does not undergo rotation and remain in fetal position by which fibulae when present lie between the tibia and the sole of the foot oriented ventrally instead of dorsal position [6]. Depending on the skeletal deformity of lower limb Sirenomelia is classified into three types–1) Apus: No foot, one tibia, one femur; 2) Unipus: one foot, two femora, two tibiae, two fibulae; 3) Dipus: two foot, two femora, two tibiae two fibulae which are fused by soft tissue giving appearance of flipper called Mermaid syndrome [2]. Stocker and Heletz has also classified Sirenomelia depending on skeletal abnormalities [Table/Fig-3] [4]. In present case the type of lower limb abnormality was of Apus type / type II.

Usually diagnosis is done in 19th-20th weeks of gestation [5,7]. In

	Characteristics
I	All thigh and leg bones present
II	Single fibula
III	Absent fibula
IV	Partially fused femurs, fused fibulae
V	Partially fused femurs
VI	Single femur, single tibia
VII	Single femur, absent tibia

[Table/Fig-3]: Shows the classification of Sirenomelia according to Stocker and Heletz [4].

present case the diagnosis was done at 28th week of gestation. Oligohydramnios occur in late pregnancy which results in poor visualization of fetal details by ultrasound in late antenatal period and diagnosis is made only after birth / at autopsy [4,5]. The differential diagnosis are caudal regression sequence, sacral agenesis, VACTERL (vertebral defects, imperforate anus, cardiac anomalies, trachea-esophageal fistula, radial and renal dysplasia, limb anomalies), Potter sequence and Fraser and Vater syndrome [3,6]. Different authors has different opinion regarding association of Sirenomelia and caudal regression sequence. Some state that the two entities are the same spectrum of the disease while others are of opinion that caudal regression syndrome sequence is pathogenetically different which has strong association with Diabetes Mellitus having normal amniotic fluid, mild deformities and heterogenous regarding etiology and developmental pathology [3]. Antenal sonographic differentiation between the two entities is important in view of the different prognosis associated with these abnormalities [3].

Prognosis is fatal and incompatible with postnatal life because of renal agenesis an its complications [2,6]. Early diagnosis with termination of pregnancy before viability i.e., less traumatic therapeutic abortion is recommended [3,4,7]. In present case medical termination was done following ultrasound findings of intrauterine fetal death and final diagnosis was done only after autopsy. However, exceptional cases are reported where the fetus survives with minor renal abnormalities or normal kidneys. However, agenesis of genitalia and anorectal atresia are invariably found [3,4,6]. Recurrence risk is unknown / not increased [6,12].

CONCLUSION

Sirenomelia is a rare lethal fetal malformation. Women during pregnancy should undergo regular antenatal checkup for early detection of fetal malformation like Sirenomelia by which the women can undergo less traumatic medical termination of pregnancy. Couples with previous pregnancy of Sirenomelia can optimistically plan future pregnancy as recurrence risk is unknown.

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