INTRODUCTION

Sirenomelia is a very rare congenital malformation with an incidence between 1:6000 and 1:100000 births. It is a lethal condition characterized by fusion of the lower limbs, single umbilical artery, and severe malformations of the urogenital and lower gastrointestinal tract. Exceptional cases of surviving newborns with minor renal abnormalities or even normal kidneys have been reported. Anomalies associated with sirenomelia in order of frequency are facial anomalies (potter face), single umbilical artery (usually the right), imperforate anus, genito-urinary system agenesis, no external genitalia or ambiguity, lower sacral/vertebral defects, cardiac anomalies, abdominal wall defects, malformed thorax and usually severe oligo/anhydramnios. The present report is a case of sirenomelia diagnosed postnatally on the basis of morphological and radiographic features. Ante-natal diagnosis was missed due to anhydramnios.

CASE REPORT

A 20-year-old primigravida presented to the obstetric outpatient department at 34 weeks of gestation. The fundal height was lower than expected for the gestational age. Her laboratory investigations were within normal limits. Her past history was unremarkable. Pre-natal ultrasound showed a breech-presenting fetus with complete absence of amniotic fluid. Adequate evaluation of the fetal morphology and anomalies was difficult due to anhydramnios, breech presentation and absent fetal movements. However, fetal cardiac activity was present. Fetal brain and visible spine were normal. Left kidney showed a single, 14.8 mm diameter cyst at its antero-medial aspect (Figure 1). The kidney showed normal size and echotexture. There was no hydronephrosis.

Right kidney was obscured by acoustic shadows from the fetal spine. Urinary bladder, caudal spine and femora could not be evaluated again due to fetal presentation. Spectral doppler trace from the umbilical artery showed a resistive index (RI) of 0.76, which suggested increased vascular resistance in the placental bed.

Elective caesarian section was performed at 35 weeks of gestation. A baby with 2.5 kg weight was delivered. Apgar score was 2 at 1, 5 and 10 minutes. The child died 2 hours after birth. Morphologically the baby had ambiguous genitalia, imperforate anus and a single, midline lower limb with absent feet and tapered distal end, giving a mermaid-like appearance. The face, the trunk and the upper extremities were normal. The radiographs of the child (Figure 2a and b) showed multiple bony abnormalities. The thorax and spine included fused transverse processes at the upper cervical, lower thoracic and upper lumbar vertebral levels, a hemivertebra and absent right pedicles of second and third lumbar vertebrae. Three right lower and two left lower ribs showed fused posterior ends. A supernumerary rib was seen on left side. Thoracic cage was small and the lungs were hypoplastic. Sacrum was hypoplastic and the coccyx was absent. Lower limb showed fusion of the femora resulting in a single midline femur articulating with the iliac bones superiorly. Tibiae were also fused. Fibulae and the bones of feet were absent. Bones of the upper limbs, scapulae and the clavicles were normal. There was no gas in the stomach or GIT suggesting esophageal atresia.

ABSTRACT

A case of sirenomelia is presented which was diagnosed on the basis of postnatal morphological and radiographic findings. The patient had single lower limb and the feet were absent. There was anal atresia and absent external genitalia. Pre-natal ultrasound showed a cyst in the left kidney and high resistive index (RI) of the fetal umbilical artery along with anhydramnios. Recent literature describes and discusses, the ante-natal diagnostic investigations.

Key words: Sirenomelia. Pre-natal diagnosis. Ultrasound. Magnetic resonance imaging (MRI).

CASE REPORT

Figure 1: Pre-natal sonographic image at 34 weeks of gestation. Left sided renal cyst is visible. Note the normal fetal brain.

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Sirenomelia has been reported as a result of teratogenic agents like, cadmium, lead, retinoic acid, and radiation. There is no widespread consensus as to the autonomous status of sirenomelia versus caudal regression syndrome (CRS), in which the dysmorphic and maldeveloped lower limbs are found in association with genito-urinary and anorectal anomalies and a variable degree of lumbosacral agenesis.\(^1\)

There is currently, no known serum marker that may be used for the ante-natal diagnosis of sirenomelia.\(^2\) Prenatal diagnosis of sirenomelia can be accomplished by sonography. In the setting of oligohydramnios and bilateral renal agenesis, the observation of lower fetal extremity fusion is the key to the ante-natal diagnosis.\(^2\) High resolution sonogram can demonstrate continuous skin line over both femurs on a transverse scan of the thigh indicating fused thighs. The echogenic skin line between the two thighs expected in separate thighs, would be absent. Alternatively single femur may be identified. However, a confident diagnosis is usually difficult during the 2nd and 3rd trimesters because of the severe oligohydramnios associated with this condition, which hampers proper evaluation of the lower extremities. This can be overcome by amnioinfusion, but it is an invasive method.\(^6\) Danilo et al. have described a case of sirenomelia with bilateral polycystic kidneys.\(^7\) The present case showed a single large cyst in the left kidney. Other sonographic features which may be apparent include; bilateral renal agenesis, absence of the bladder, undetermined external genitalia, anorectal atresia and lumbosacral agenesis. Other frequent abnormalities involve the abdominal wall and cardiovascular system. Hypertrophy of the right ventricle may ensue in later stages as a consequence of renal agenesis and oligohydramnios. Doppler flow imaging invariably reveals a two-vessel umbilical cord.\(^1\)

Alternatively, magnetic resonance imaging (MRI) can be used for the ante-natal diagnosis, because it is less hampered by oligohydramnios, but it is not easily available.\(^6\) In addition, the safety of MRI in pregnancy is not widely established. The use of MRI is, therefore, recommended only in case of strong suspicion of the fetal anomaly which would subsequently need termination of pregnancy.

It is important to establish correctly the pre-natal diagnosis of sirenomelia or CRS in a fetus with dysmorphic lower limbs, because of different prognoses associated with these two conditions. Termination of pregnancy is indicated in sirenomelia and not in CRS. In CRS, the prognosis depends upon the extent of spinal abnormalities.\(^4\)

It is, therefore, recommended to maintain a high index of suspicion for sirenomelia in patients with severe oligo/ anhydramnios. Importance of early second trimester anomaly scan is also stressed for the early and easy diagnosis of sirenomelia.

**REFERENCES**


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**DISCUSSION**

Dysmorphic lower limbs are the principal feature of sirenomelia.\(^1\) Stocker et al. have classified sirenomelia into seven variants, ranging from fusion of soft tissue to a single limb (Table I).\(^3\) The lower limbs may be fused to the extent of giving a mermaid-like appearance, hence the name mermaid malformation.\(^4\) The patient in the present case was identified as type-VI (single femur, single tibia) according to this classification. Akhtar et al. have reported a case of sirenomelia in which the baby had 2 femoral and 2 tibial bones.\(^5\) Fibulae and the bones of feet were absent.

<table>
<thead>
<tr>
<th>Type</th>
<th>Characteristics</th>
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<tbody>
<tr>
<td>I</td>
<td>All thigh and leg bones are present</td>
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<tr>
<td>II</td>
<td>Single fibula</td>
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<tr>
<td>III</td>
<td>Absent fibula</td>
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<tr>
<td>IV</td>
<td>Partially fused femurs, fused fibulae</td>
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<tr>
<td>V</td>
<td>Partially fused femurs</td>
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<tr>
<td>VI</td>
<td>Single femur, single tibia</td>
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<tr>
<td>VII</td>
<td>Single femur, absent tibia</td>
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Table I: Classification of sirenomelia.\(^3\)

**Figure 2:** (a) Frontal and (b) lateral radiographs of the infant showing femoral and tibial fusion. Sacrum, fibulae and bones of the feet are absent. Multiple abnormalities of the spine and ribs are also visible.