PRESENTATION OF CASE

We are presenting two cases of Sirenomelia. It derives its name by its resemblance to the Mermaid (Siren) of Greek mythology and called as Mermaid syndrome. It is a rare congenital anomaly which presents with variable degree of fusion of lower limbs, sacral and pelvic bony anomalies, absence of external genitalia, imperforate anus and renal agenesis or dysgenesis. There are approximately 300 cases reported in the literature, 15% of which are associated with twinning, most often monozygotic. Caudal Regression Syndrome is thought to be the result of injury to the caudal mesoderm early in gestation. Agenesis of left kidney was found in one of our cases on autopsy.

Case I - A 21-year-old pregnant female with 19 weeks of gestation was referred to our hospital in view of anomalous foetus. Obstetric ultrasound scan at Rajarajeswari Medical College and Hospital, Bengaluru, revealed multiple anomalies for which termination of pregnancy was advised by the obstetrician after counseling.

Post expulsion, physical examination of baby revealed both legs were joined up to the thigh with bilateral patellae facing posteriorly (Figure 1), absence of anal orifice and intact foetal spine.

Infantogram performed on the baby (Figure 2) showed dysplastic bilateral pelvic bones with mild malrotation, soft tissue fusion of proximal lower limbs with agenesis of bilateral fibula and feet with normal foetal spine and visualized other bones. Both the femurs were normal in size for the baby age.

Figure 2. AP and Lateral views of Infantogram Bilateral Dysplastic Pelvic Bones with Soft Tissue Fusion of Proximal Lower Limbs and Agenesis of Bilateral Fibula and Feet

Case II - A 30-year-old pregnant woman presented to our hospital in active labour. Clinical examination revealed approximate gestational age of 32 weeks. Ultrasound scan was not done previously. Since the new born baby didn't cry immediately after birth and required resuscitation was not done in view of multiple anomalies and baby was kept under hood oxygen. The neonate did not survive this lethal abnormality and died after 4 and half hours.

Physical examination revealed fusion of lower limbs, absence of anal orifice and non-development of external genitalia (Figure 3).

Infantogram showed agenesis of fifth lumbar vertebrae, unfused hip bone and agenesis of sacrum and coccyx, single femur not articulating with hip bone and single tibia and narrow upper part of the chest. Cervical vertebrae were normal (Figure 4).

Autopsy performed on the foetus showed multiple anomalies like collapsed foetal lungs, pyloric part of stomach continuing as duodenum with a blind end diverticulum filled with meconium, small and large intestines with blind ends at the beginning and termination and agenesis of left kidney. Heart, liver and pancreas were normal (Figure 5).
CLINICORADIOLOGICAL DIAGNOSIS

Based on the characteristic clinical and radiological findings both the babies were diagnosed to have Sirenomelia. Based on the presence or absence of bones and Stocker and Heifetz classification both cases were categorized into Simpus dipus and Type –III and Simpus apus and Type –VI respectively.

DISCUSSION OF MANAGEMENT

Sirenomelia is rare and fatal congenital anomaly with an incidence of 0.8 to 1 case per 1,00,000 births and Male to Female ratio of 3:1. The first medical description of Sirenomelia was given by Rocheus and Polfyr in the sixteenth century.¹

Duhamel et al considered this anomaly as a manifestation of the caudal regression syndrome which is a consequence of abnormal development of structures derived from the caudal mesoderm of the embryo before the fourth week of gestation and extended to various cranio-caudal levels.² Formation of the lower vertebral column and corresponding nervous system is usually completed by 4th week of development. Owing to abnormal gastrulation, the mesoderm migration is disturbed. This disturbance may result in lesions varying from minor changes in vertebrae to complete fusion of the lower limbs.³ The embryological defect occurs at the mid-posterior axis of the mesoderm leading to arrest of progression of the mesoblastic caudal bud. Studies of axial mesoderm patterning at early gestation suggest that one or more processes of primitive streak migration, neutralisation or differentiation are compromised.⁴ According to theory of multisegmental early embryonic chorda-mesodermal derangement, severity of the malformation and its segmental level would affect the residual spinal function and the clinical deficit. If the derangement involves the most caudal segment, it will result with caudal regression syndrome.⁵

Though the exact aetipathogenesis is uncertain, the most accepted one was vascular steal or vitelline artery steal theory by Stevenson et al. The study of the eleven Sirenomelia specimens showed, a large artery arose from the aorta high in the abdomen beyond which the aorta and its branches were hypoplastic denying the blood and nutrients to the caudal part of the embryo.⁶

Maternal diabetes is the main risk factor for caudal regression syndrome occurring with greater frequency in women with diabetes. The relative risk for developing this syndrome is 1:200 to 1:250 with maternal diabetes and 22% of foetuses with this anomaly will have diabetic mothers.⁷ Other environmental risk factors are smoking, heavy metal exposure and retinoic acid.⁸

The characteristic finding of Sirenomelia is partial or complete fusion of the lower legs. There is highly variable degree of severity. Affected infants may have only one femur or may have two femurs within one shaft of the skin. In some affected infants there may be one foot, no feet or both feet, which may be rotated so that the back of the foot is facing forward. The affected infants may also have a unilateral or bilateral agenesis, cystic malformation of the
kidneys, absence of bladder, urethra, urethral atresia. In addition, imperforate anus may also be seen. Infants with sirenomelia may also have abnormalities affecting the sacral and lumbar spine.

**Classifications**

I. Sirenomelia has been classified into three types according to the number of lower limb bones present.¹

<table>
<thead>
<tr>
<th>Type</th>
<th>Femur</th>
<th>Tibia</th>
<th>Fibula</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>I</td>
<td>2</td>
<td>2</td>
<td>2</td>
<td>All bones present</td>
</tr>
<tr>
<td>II</td>
<td>2</td>
<td>2</td>
<td>1</td>
<td>Single fused fibula</td>
</tr>
<tr>
<td>III</td>
<td>2</td>
<td>2</td>
<td>0</td>
<td>Fibula absent</td>
</tr>
<tr>
<td>IV</td>
<td></td>
<td></td>
<td></td>
<td>Partially fused</td>
</tr>
<tr>
<td>V</td>
<td></td>
<td></td>
<td></td>
<td>Partially fused; fibula fused</td>
</tr>
<tr>
<td>VI</td>
<td>1</td>
<td>1</td>
<td>NA</td>
<td>Single femur; single tibia</td>
</tr>
<tr>
<td>VII</td>
<td>1</td>
<td>0</td>
<td>NA</td>
<td>Single femur; tibia absent</td>
</tr>
</tbody>
</table>

Stocker and Heifetz classified Sirenomelial infants from Type I to Type VII according to the presence or absence of bones within the lower limb.¹¹

Prenatal ultrasonography as early as 13 weeks of pregnancy can detect gross structural anomalies. So, if diagnosed early the alternative of termination of pregnancy can be safely advised to the mother. Proper control of blood glucose level in a diabetic mother may prevent the occurrence of sirenomelia.⁷

**FINAL DIAGNOSIS**

The first case is simpus dipus according to first classification and type III according to Stocker and Heifetz.

The second case is simpus apus according to first classification and type VI according to Stocker and Heifetz.

**CONCLUSION**

Sirenomelia is a rare and lethal congenital anomaly. When diagnosed antenatally, termination should be offered. However, prevention is possible and should be the goal. Regular antenatal checkup with optimum maternal blood glucose level in preconception period and in first trimester should be maintained to prevent this anomaly.

**REFERENCES**