A Rare Association of Sirenomelia (Symelia Unipus) with Isolated Levocardia

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ABSTRACT: Sirenomelia or the “mermaid syndrome” is a rare entity. Malformations of almost every system have been reported in sirenomelia and it is invariably incompatible with survival; most babies are stillborn, or die shortly after birth. Isolated levocardia is an extremely rare condition in which the heart is located in the normal position while abdominal viscera are inverted. We report a case of symelia unipus having situs inversus of abdominal viscera with isolated levocardia, a ventricular septal defect, bilateral genitourinary agenesis and thoracic vertebral defects.

KEYWORDS: Mermaid syndrome, isolated levocardia, sirenomelia, situs inversus

Sirenomelia or ‘mermaid syndrome’ is a rare and lethal malformation with a reported incidence of up to 1 in 100,000 live births. The term hails from “siren” or "mermaid" of Greek mythology. It is characterized by single or fused lower limbs, associated with other anomalies of lumbosacral and pelvic bone, blind colon, absent external genitalia, single umbilical artery and renal agenesis. It is considered the severest form of caudal regression syndrome, a congenital defect with variable spinal anomalies ranging from partial sacral agenesis to complete absence of the lumbosacral spine. The prenatal diagnosis is severely limited by the accompanying oligohydramnios that hinders visualization of fetal anatomy. About 300 cases have been reported in literature of which 15 have been reported from India. Only 6 cases have been reported to survive the neonatal period.

Case Report

A 24-year-old non-diabetic primigravida mother delivered a singleton baby at 36 weeks’ of gestation by spontaneous vaginal delivery. There was no history of consanguinity, drug or radiation exposure or any other illness during pregnancy. Antenatal ultrasound examination, done two days prior to delivery revealed severe oligohydramnios with non visualization of the fetal renal structures.

At birth the baby had cyanosis with gasping respiration and needed positive pressure ventilation. One minute Apgar score was 4 and five minute score was 7. On examination both the lower limbs of the baby were fused to a single mass ending in a single foot which showed reversal of the normal heel-toe anteroposterior orientation and a single phalanx (Fig.1).
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There was no external genitalia, no urethral or anal opening. The upper torso of the baby was normal. No facial dysmorphism was evident. Weight of the baby was 1380 g, length was 34 cm and head circumference was 28.7 cm. The baby died at 3 hours of age.

Postmortem radiograph (Fig.2) showed complete sacrococcygeal agenesis, hypoplastic iliac bones, fused lower extremities with paired femur and tibia, absent fibulae and a fused metacarpal bone in the rudimentary foot. Hemivertebra of T10 vertebrae with fusion of T11 and T12 vertebrae and marked lower thoracic kyphoscoliosis were noted.

Autopsy revealed a severely contracted pelvic cavity with bilateral agenesis of kidneys, gonads, genitourinary tract and adrenals. The liver was left sided with stomach and spleen on the right side. The anus and rectum were absent with a blindly terminating rectosigmoid. Two umbilical arteries were found arising normally from the iliac arteries in continuity with the abdominal aorta. Both lungs were hypoplastic. The heart was normally oriented on the left side with a membranous ventricular septal defect. Karyotyping could not be carried out.

**Results and Discussions**

In sirenomelia, fusion of the lower extremities can range from membranous fusion of soft tissue to total fusion of the lower limb with one midline femur. Sirenomelia is classified according to the number of bones in the lower limb. The baby we described was symelia unipus, type III variant (absent fibulae) according to the classification by Stocker and Heifetz. Additionally this baby also had severe lower thoracic kyphoscoliosis.
with hemivertebrae and fusion of T11 and T 12 vertebral bodies.

A male/female ratio of 2.7:1 is reported but because there were no external genitalia or gonads and since we could not perform karyotyping the sex of this baby was not ascertained. Renal dysplasia is found to be almost always present in sirenomelia (> 90%). Sirenomelia is lethal due to bilateral renal agenesis leading to severe oligohydramnios and lung hypoplasia. Most infants are stillborn or die a few hours after birth. In addition to renal agenesis and lung hypoplasia the entire genitourinary system and adrenals were absent in this baby.

Several hypotheses for the etiology of the sirenomelia sequence have been proposed. Duhamel suggested that it was a part of the caudal regression sequence. The most popular hypothesis is the “vitelline artery steal” theory by Stevenson et al who inferred that sirenomelia results because a single umbilical artery arising high on the abdominal aorta diverts blood away from the caudal portion of the embryo to the placenta. Another theory suggests that disruption of caudal mesoderm during the gastrulation stage in the 3rd week of gestation interferes with formation of the notochord causing excessive primary cell death in the caudal mesoderm and hindgut endoderm leads to abnormal caudal structures. Langer et al in their report of a case of sirenomelia associated with situs inversus proposed that a defect in the midline blastogenesis might cause the sirenomelia sequence. In up to 80% of cases a single umbilical artery has been found unlike in this baby where there were two normally arising umbilical arteries. This leads to speculation that the vitelline artery steal phenomenon does not hold true in this case.

Isolated levocardia is an extremely rare type of situs inversus in which the heart is in the normal levo position, but the abdominal viscera are in the dextro position. There is typically a right arch with complex cardiac defects. Isolated levocardia is distinct from heterotaxy syndrome where lung isomerism (left or right) is found. Stanton et al reported dextrocardia and situs solitus in a case of sirenomelia. Our case had situs inversus of the abdominal viscera with isolated levocardia apart from two normal umbilical arteries. These findings support the theory forwarded by Langer et al. There is an apparent overlap between the phenotypic manifestations of sirenomelia and the VACTERL complex. Stocker and Heifetz documented at least three of the components of VACTERL in all 80 cases of sirenomelia. The present case had 6 of the components of VACTERL, reinforcing the theories that axial mesodermal defects or the spectrum of caudal dysgenesis i.e. factors other than vitelline artery steal produce sirenomelia.

Sirenomelia carries a grim prognosis. Diagnosis is often missed as during the late second and third trimesters, severe oligohydramnios or anhydramnios may limit the diagnosis of sirenomelia in ultrasonography. Survival depends on the existence of adequate kidney function. Till date there is no definite evidence of a genetic basis for sirenomelia sequence.

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