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Sirenomelia: Mermaid deformity on fetal MR imaging

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This case involves the rare congenital disorder Sirenomelia, a diagnosis initially suspected during prenatal ultrasound and later confirmed by prenatal MRI. Sirenomelia, or mermaid syndrome, is mainly characterized by variable fusion of the lower limbs and by genitourinary anomalies. The vast majority of cases of this disease result in death secondary to associated renal agenesis or hypoplasia. Imaging findings from the disease are discussed, in addition to the clinical implications and suspected etiologies of this entity.

Case report

Fetal MR imaging was performed on a 24-week (gestational age) female to further investigate multiple anomalies identified on prenatal sonogram. The amniotic fluid level was noted to be low normal, and the kidneys were suspected to be hypoplastic with absence of the urinary bladder (Fig. 1). Examination of the heart demonstrated cardiomegaly (Figs. 2, 4, and 5). The umbilical vein bypassed the liver, crossing the liver surface to enter the right atrium (Fig. 4). There was abnormal curvature and positioning of the lower limbs, which appeared fused (Figs. 1 and 3). Lumbar hemivertebra was observed with resulting kyphotic deformity of the spine (Fig. 1). Atrial and ventricular septal defects were detected on prior prenatal sonogram but were not apparent on the prenatal MRI.

The child was later born via uncomplicated spontaneous vaginal delivery at 35 weeks, 4 days, to a 22-year-old primigravida. The child's mother admitted to smoking early in the pregnancy, but denied any other drug use or significant past medical history. On physical examination, the child had a two-vessel umbilical cord, absent anus and genitalia, and a sacral dimple. There was fusion of the lower extremities from the hips to the feet (Figs. 6, 7). Postnatal cardiac ultrasound demeonstrated a double-outlet right ventricle, mitral-valve hypoplasia, patent foramen ovale, moderate obstruction of the pulmonary-outflow tract, large malalignment-type ventricular septal defect, and mild left-ventricular hypoplasia.

The child required intubation soon after birth due to respiratory distress. Postnatal renal ultrasound confirmed absence of the kidneys and urinary bladder, and the patient failed an intravenous fluid challenge. Care was withdrawn as per the family's wishes, and the child died two days after birth.

Discussion

Sirenomelia, or "mermaid deformity," is a rare congenital disease sequence with an incidence of approximately 1 in 100,000 births (1). Sirenomelia is characterized by maldevelopment of the caudal portions of the body, with genitourinary abnormalities and varying degrees of fusion of the lower extremities. The disorder is classified into three types according to the number of feet: simpus apus, no feet present; simpus unipus, one foot; simpus dipus, two feet (2). Complete or partial bilateral renal agenesis is common and most often incompatible with life, although there have been very rare reported cases of patient survival secondary to the presence of a normal kidney. The absence of genitalia and anorectal anomalies are also invariably encountered.

The exact etiology of sirenomelia remains incompletely understood. A popular theory is abnormal persistence of the vitelline artery, leading to vascular steal from the abdominal aorta and lower extremity. Other suggested etiologies include teratogenic agents, maternal diabetes, and ge-

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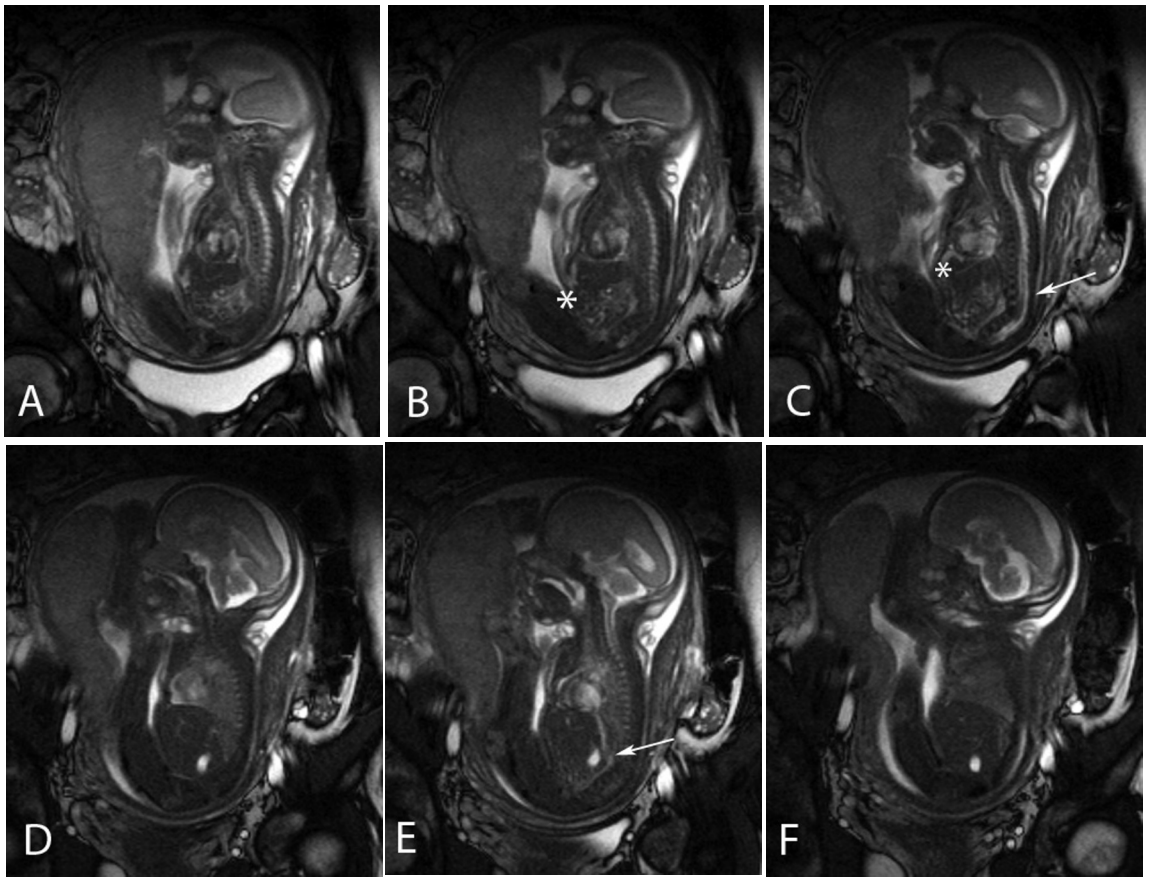

Figure 1. 24-week-old fetal female with sirenomelia. Sagittal T2 MR images demonstrate fused lower extremities (D), absent kidneys, absent urinary bladder, lumbar hemivertebra resulting in focal kyphosis (C, arrow), and oligohydramnios. Abnormal high insertion of the umbilical vein is present (B, C; star). Gallbladder is also noted (E, arrow).

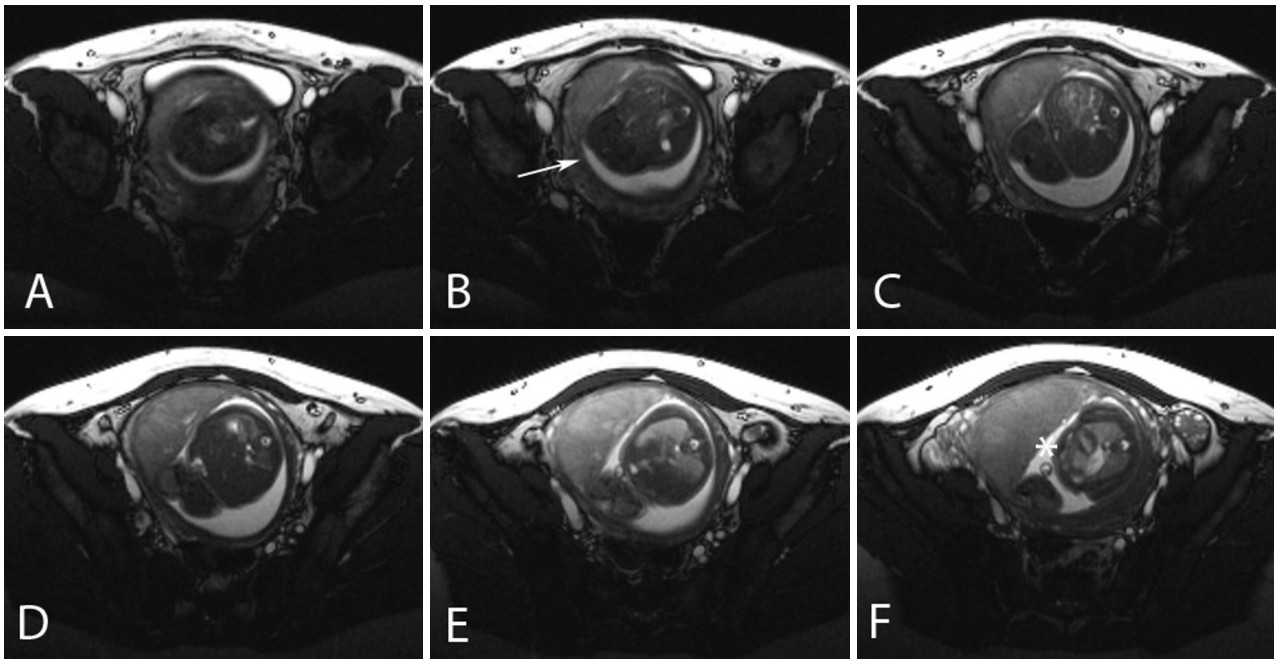

Figure 2. 24-week-old fetal female with sirenomelia. Axial T2 MR images demonstrate absent urinary bladder (A), fused lower extremities (B-F), absent kidneys (B, C), cardiomegaly (F), oligohydramnios, and two-vessel umbilical cord (E, F [star]).

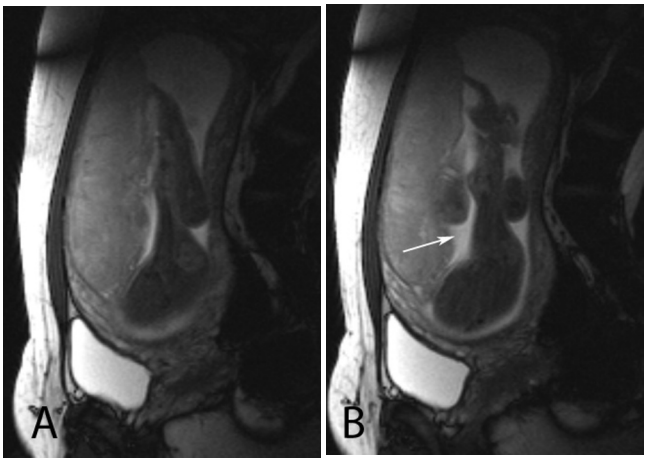

Figure 3. 24-week-old fetal female with sirenomelia. Coronal T2 MR images demonstrate fused, abnormally positioned, fixed, and extended lower extremities (arrow).

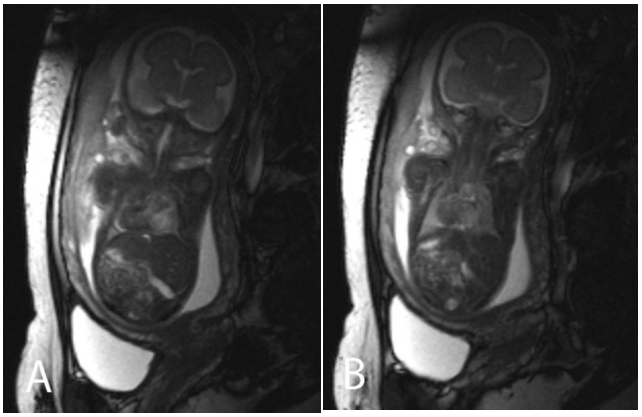

Figure 5. 24-week-old fetal female with sirenomelia. T2 coronal images demonstrate cardiomegaly (A) and hypoplastic lungs (B).

netic predisposition. Some suggest that sirenomelia is related to the caudal regression syndrome (CRS), as many of the anomalous features are similar (1). However, the presence of a single umbilical artery in sirenomelia seems to separate the two entities, as this is not a feature of CRS.

Sonographic evaluation of the fetus in the second and third trimesters is usually limited secondary to severe oligohydramnios from renal agenesis. However, diagnosis can be suggested by visualization of a fused femur, decreased distance between two femurs, or decreased/absent motility of the lower extremities (2). A single umbilical artery is often seen. Other sonographically apparent features include absent urinary bladder, undetermined genitalia, lumbosacral agenesis, and anorectal atresia. Prenatal MRI can easily demonstrate the above findings, without the constraints of oligohydramnios.

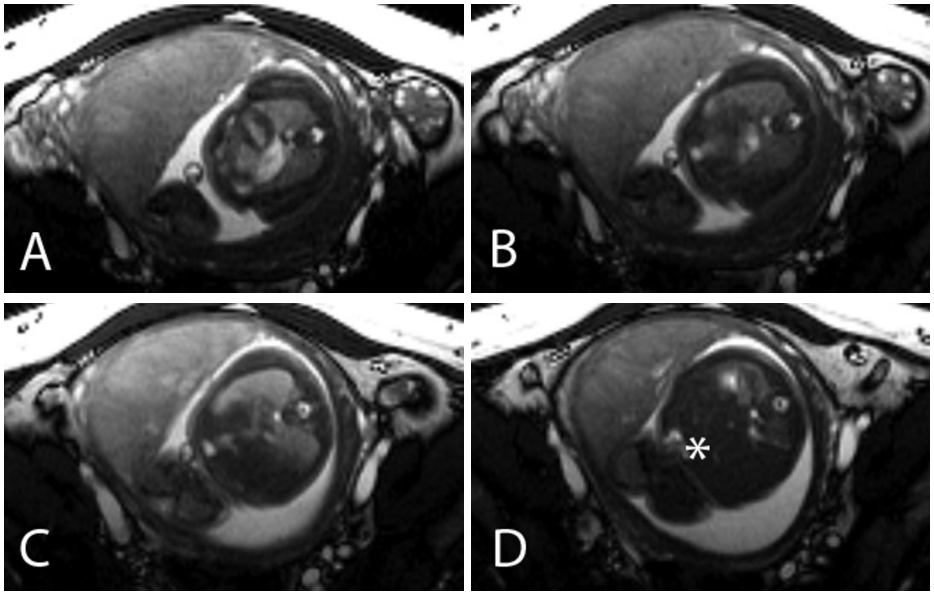

Figure 4. 24-week-old fetal female with sirenomelia. Axial T2-weighted images demonstrate two vessel umbilical cord (A-C) and abnormal high insertion of the umbilical vein draining into right atrium (D, star).

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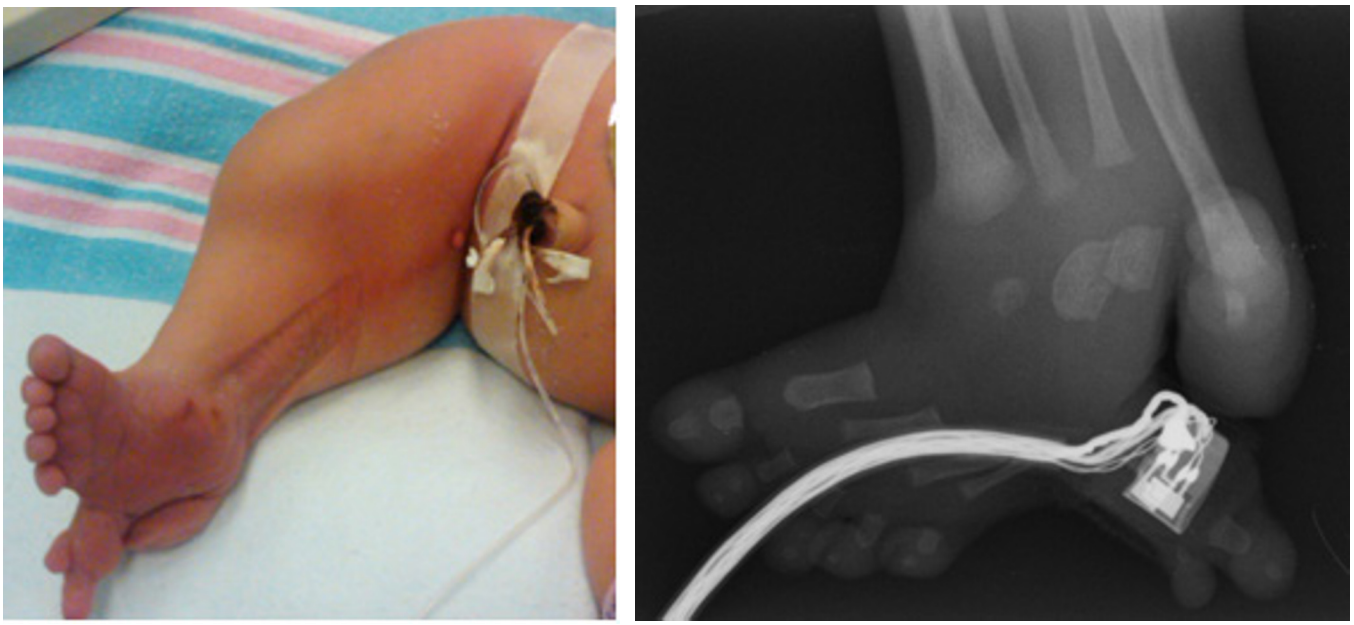

Figure 6. Infant female with sirenomelia. Postpartum photo (left) and radiograph (right) demonstrate fused and abnormally positioned legs with flipper/mermaid deformity.

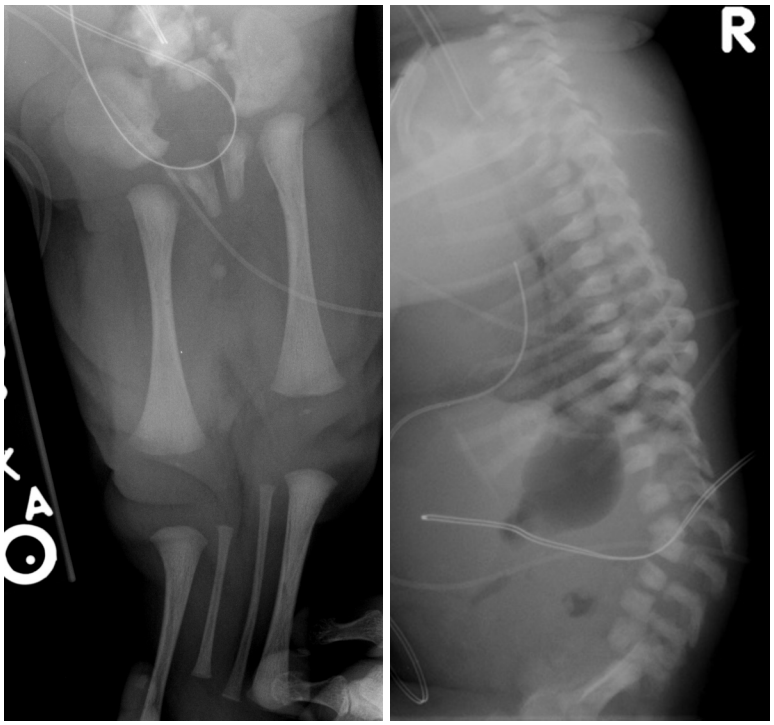

Figure 7. Infant female with sirenomelia. Postpartum radiographs redemonstrate the spinal and extremity deformities.