A case of postnatally diagnosed Sirenomelia in a patient with poorly controlled type 2 diabetes mellitus

Background/Synopsis

Sirenomelia, or Mermaid Syndrome, is a rare congenital anomaly of caudal fetal development, causing a complete absence or fusion of lower limbs. It affects approximately 0.98 per 100,000 live births. Most newborns die within the first few hours of life. Several risk factors have been associated with Sirenomelia, including maternal hyperglycemia, young maternal age, and monozygotic twinning, but this condition's etiology and pathogenesis are poorly understood.

Objectives/Purpose

We present a case of a 34-year-old with a history of poorly controlled type 2 diabetes mellitus and anhydramnios at 17 weeks gestation. She was managed as presumed pre-viable premature rupture of membranes. However, she likely had renal agenesis resulting in anhydramnios; Sirenomelia with renal agenesis was diagnosed after delivery, illustrating the importance of earlier anatomy ultrasound in high-risk patients.

Case Report

A 34 yr old G5P3013 presented for consultation due to poorly controlled T2DM at 13.6wga by ultrasound not c/w LMP. She was diagnosed with pre-viable PPROM after an ultrasound at 17 weeks revealed new onset anhydramnios with severe fetal growth restriction and multiple congenital anomalies.

The patient reported leaking fluid for weeks, but a physical exam revealed no pooling or ferning, with a negative nitrazine test. She was counseled on the poor prognosis but desired aggressive management. She received latency antibiotics. Follow-up ultrasound showed suspected holoprosencephaly, AV canal defect, unilateral renal agenesis, and abnormal cervical and lumbar spine curvature.

She reiterated her desire for all fetal interventions with pediatric palliative care. She was hospitalized at 23.0wga for BMZ and MgS04 and met with the NICU team, who reiterated that prognosis was likely poor. Her glucose control continued to be poor secondary to diet and the patient’s frequent insulin refusal. She remained inpatient until a repeat cesarean section at 34.0wga.

At delivery, the fetus had APGARS of 2 and 8 at one and five minutes, respectively, with a fetal weight of 1240gm. Physical exam demonstrated microcephaly with a defect in the cranial bones, asymmetric chest, two-vessel cord, ambiguous genitalia, imperforate anus, webbed legs with knee joints fused bilaterally, fused feet with 11 total toes, no primitive reflexes, and no palpable femoral pulses. In the NICU, CXR showed bilateral pneumothoraxes requiring thoracenteses. The abdominal US showed renal agenesis without fluid in the bladder, and a decision was made for no further interventions. The infant later experienced cardiac arrest on the first day of life.

Microarray, whole exome sequencing, and mitochondrial sequencing were all negative. She was counseled that the anomaly was most likely secondary to poor glucose control and recommended to achieve better control before her subsequent pregnancy if one was desired.

Conclusion

Sirenomelia remains a fatal condition with multisystem involvement. While there are reports of a few surviving neonates, these babies generally have had poor outcomes. This condition is difficult to diagnose in the second trimester due to anhydramnios obscuring visualization of anatomic structures. ISUOG has guidelines for the assessment of fetal anatomy in the first trimester. In obese diabetics, the 11-13 weeks TVUS may represent the best opportunity to evaluate for diabetic fetopathy. It would likely have revealed the defects seen in this case.

Sirenomelia was previously considered a severe form of caudal regression syndrome but is now recognized as a separate disease. While the exact etiology is unknown, it's likely secondary to genetic and environmental factors that occur before embryogenesis leading to compromised vasculature formation in the lower body. While most cases appear sporadic, poor glucose control prior to conception is a known risk factor, and women with pre-existing diabetes should seek preconception counseling to reduce the risk of poor pregnancy outcomes.