

First Trimester Diagnosis of Suspected Volvulus and Sirenomelia: A Rare Lethal Association

Dr Bhawna Garg

Introduction

- Sirenomelia is a rare, lethal congenital anomaly characterized by complete or incomplete fusion of the lower limbs.
- Frequently associated with renal agenesis, severe oligohydramnios, blind-ending gastrointestinal tract, and single vitelline artery.
- The concurrent presentation of sirenomelia with midgut malrotation and active volvulus is exceptionally rare.
- Detecting this complex multi-systemic malformation during early first-trimester screening poses significant diagnostic and technical challenges.

Case Presentation

- **Patient Profile:** A 24-year-old primigravida presented for routine first-trimester aneuploidy screening at 12 weeks 3 days of gestation.
- **Transvaginal Ultrasound Findings:**
 - Single lower extremity lacking femoral bifurcation, characteristic of sirenomelia.
 - Abnormal bowel pattern displaying a classic sonographic “whirlpool sign”, highly suggestive of volvulus.
 - Colour Doppler revealed a single vitelline artery. Urinary bladder was not visualized.
- Post-mortem examination confirmed the antenatal findings following elective termination.

Discussion & Pathophysiology

The coexistence of caudal regression features and gut malrotation points toward a common early vascular insult specifically vitelline arterial steal/maldevelopment.

Early Vascular Disruption / Vitelline Vessel Anomaly	
Vascular Steal Sequelae (Caudal Ischemia) • Lower limb fusion • Renal/genital agenesis • Anorectal atresia	Midgut Vascular Compromise (SMA / Vitelline Disruption) • Defective gut rotation • Shortened, loose mesentery • High risk for Volvulus

Conclusion

✓ Diagnostic Value

High-resolution 3D transvaginal ultrasound combined with Colour Doppler imaging is an indispensable tool for diagnosing complex, rare lethal anomalies in the first trimester.

✓ Clinical Takeaway

Recognizing the syndromic association between sirenomelia and midgut volvulus enhances early prognostic assessment, ensuring comprehensive counselling for affected families.

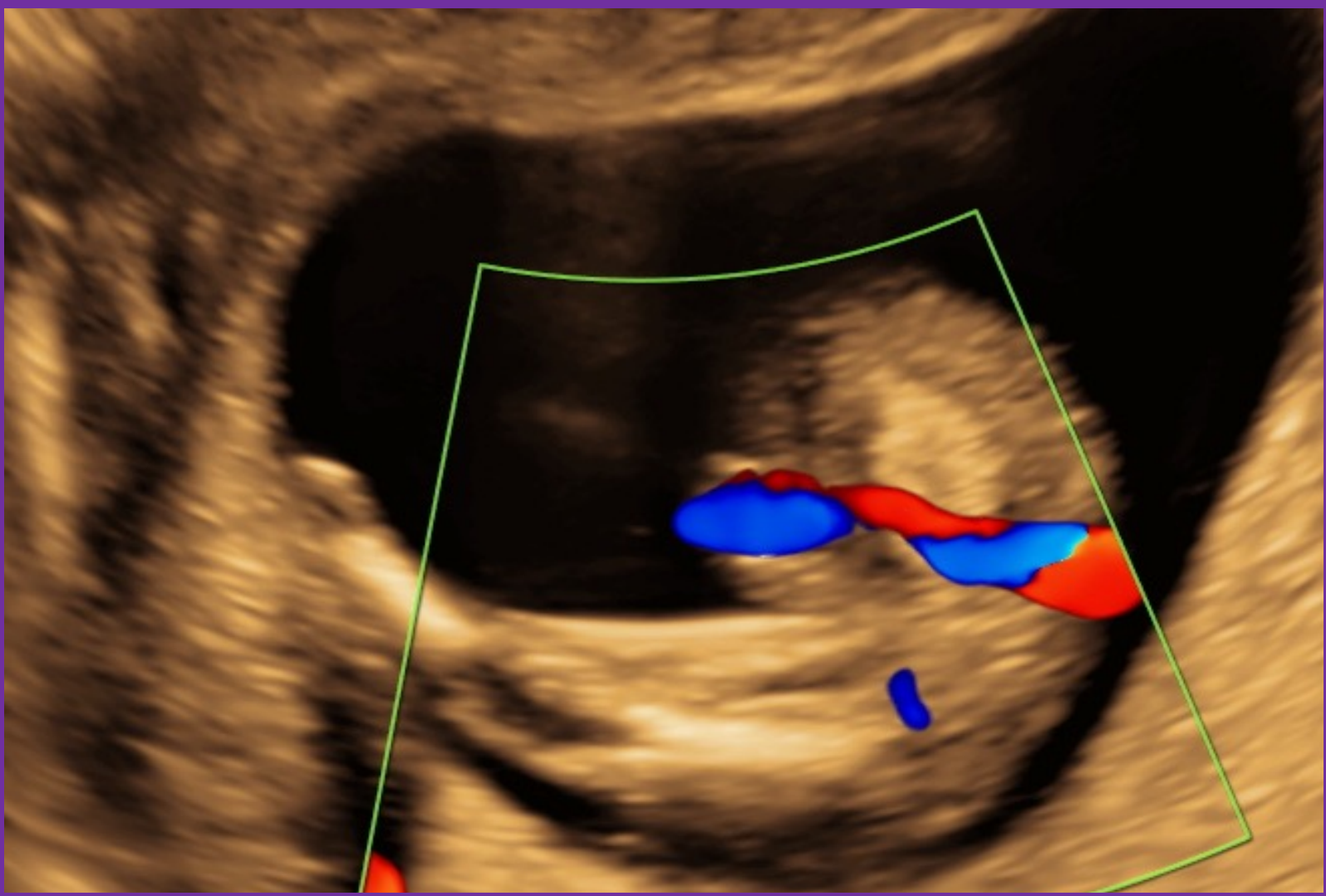


Figure 1: Single Vitelline artery



Figure 2: Single lower extremity

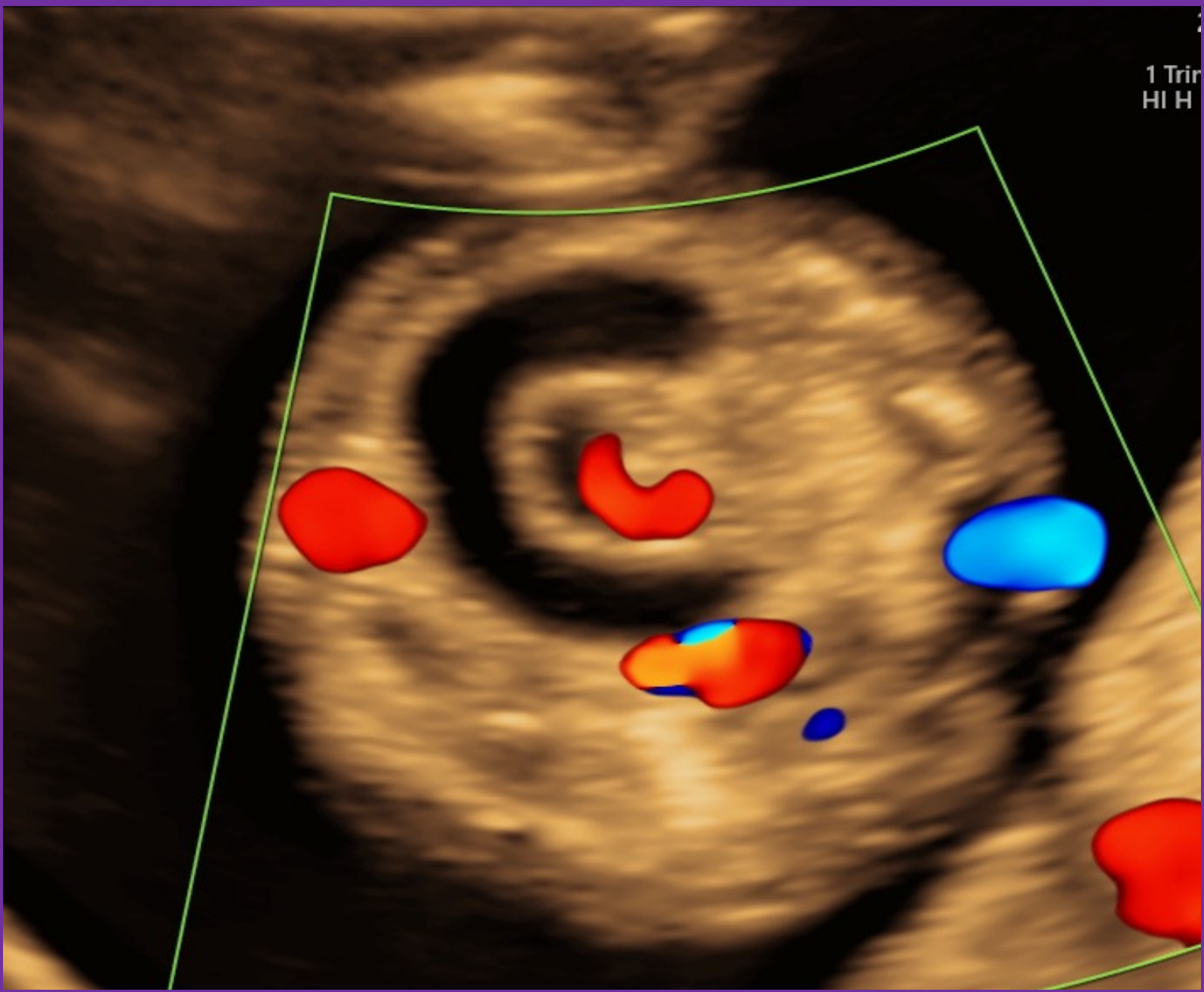


Figure 3,4: Abnormal bowel pattern displaying whirlpool sign



Figure 5: 3-D demonstration of Sirenomelia



Figure 4: Postabortal confirmation of Sirenomelia