



EP05.04 Case report: first-trimester diagnosis of situs inversus and novel CFAP53 variants



Maria-Ioanna Chatziioannou, Afroditi Pegkou, Angeliki Rouvali, Ioakeim Sapantzoglou, Antonia Varthaliti, Konstantinos Tasiaas,

Zacharias Fasoulakis, Alexandros Psarris, Athena Souka, Panos Antsaklis, Georgios Daskalakis, Marianna Theodora

1st Department of Obstetrics and Gynecology, National and Kapodistrian University of Athens, Greece

Introduction

Situs Inversus Totalis (SIT) is a rare congenital condition and involves the complete transposition of all visceral organs. Prognosis is largely determined by its association with Primary Ciliary Dyskinesia (PCD). Identifying whether the underlying genetic etiology affects only the embryonic nodal cilia or extends to postnatal respiratory cilia is critical for predicting long-term morbidity.

Case presentation

A 41-year-old primigravida presented for the nuchal translucency scan at 12 weeks of gestation. Ultrasound revealed right-sided stomach and dextrocardia. Fetal echocardiography in the early second trimester confirmed dextrocardia and a structurally normal heart. Fetal magnetic resonance imaging (MRI) confirmed SIT with the liver located on the left and normal gallbladder morphology. The large bowel was visualized to the right of the midline; therefore, postnatal contrast enema was recommended to exclude intestinal malrotation and was subsequently normal. Following amniocentesis,

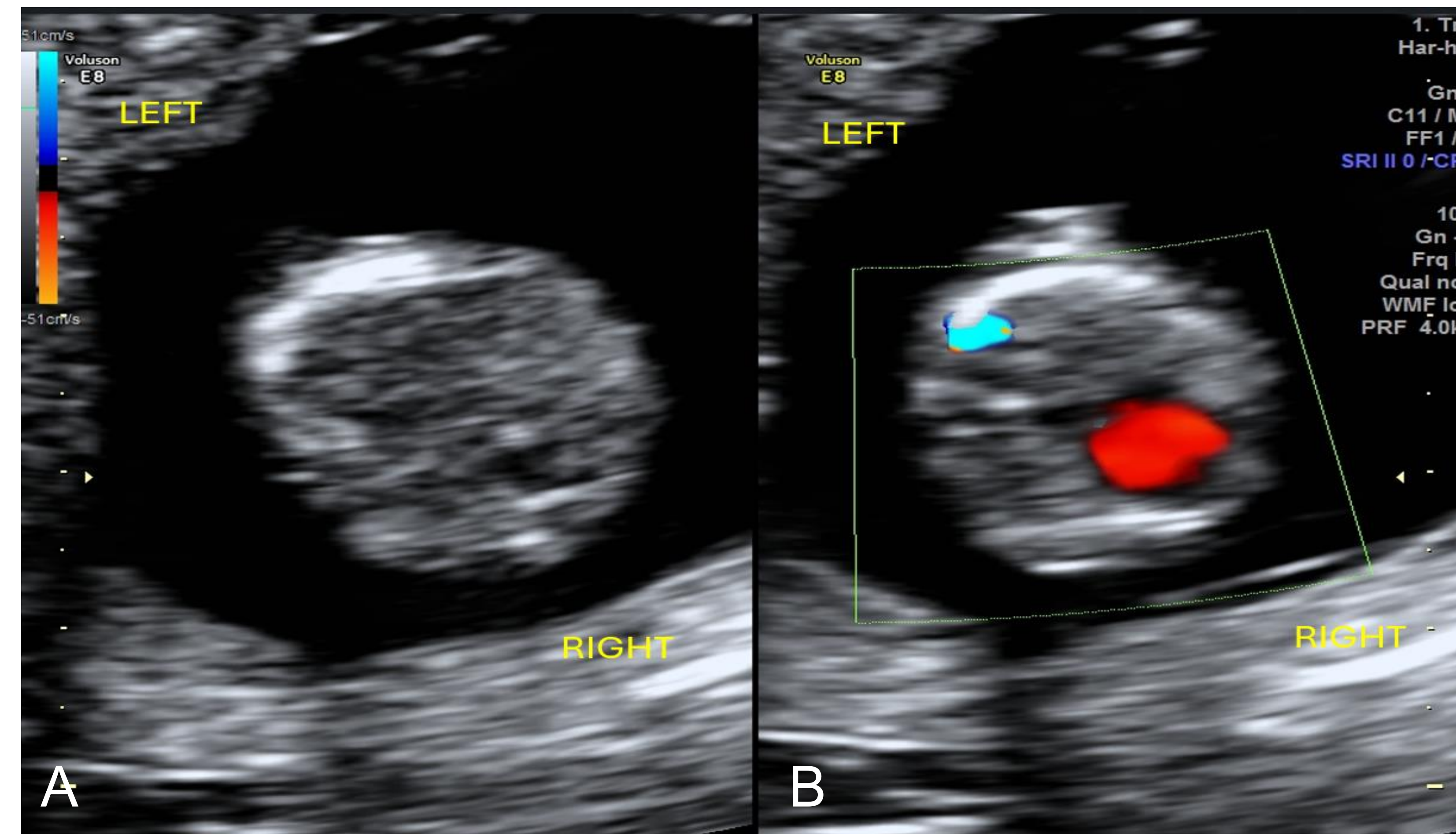


Figure. First-trimester ultrasound demonstrating situs inversus totalis. (A) Transverse abdominal view showing a right-sided stomach. (B) Four-chamber view demonstrating dextrocardia.

karyotype and chromosomal microarray analysis results were normal, however, Whole Exome Sequencing (WES) identified compound heterozygous variants in the CFAP53 gene: a frameshift deletion (c.870del) and a splice-site variant (c.473+1G>A), that were classified as likely pathogenic.

CFAP53 encodes a protein involved in the structural organization of motile cilia affecting left–right axis determination during embryogenesis. Unlike classic ciliopathies such as PCD, CFAP53-related disorders may selectively affect nodal cilia involved in early embryonic symmetry breaking while sparing respiratory cilia.

Conclusion

In this case, early detection of SIT allowed a critical window for evaluation such as fetal echocardiography and MRI to exclude heterotaxy-related anomalies. Furthermore, this case expands the spectrum of CFAP53 mutations and highlights the diagnostic value of prenatal WES in fetuses with SIT. Identifying the genetic cause allows precise counselling about prognosis and the risk of recurrence.



36TH ISUOG WORLD CONGRESS
ON ULTRASOUND IN OBSTETRICS & GYNECOLOGY
LONDON, UK 4-7 SEPTEMBER 2026

