

# Midgut Volvulus in Trisomy 21: Unveiled on Fetal Autopsy

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## Abstract

Midgut volvulus is a rare developmental anomaly with a largely unknown etiology that is challenging to diagnose antenatally in the second trimester. The objective of this case report is to describe a rare case of midgut volvulus associated with trisomy 21. Amniocentesis was done in a 19-week fetus with severe ventriculomegaly, hypoplastic cerebellum, echogenic bowel, and growth restriction to rule out chromosomal anomalies. The couple opted to terminate the pregnancy in view of the severity of the anomalies. The fetal autopsy revealed a characteristic whirlpool appearance of midgut volvulus, and the karyotype was suggestive of trisomy 21. This is the second reported case of midgut volvulus associated with trisomy 21 where ultrasound was not specific for this anomaly and the gut loops were coiled in a distinctive whirlpool pattern detected incidentally on fetal autopsy.

**Keywords:** Midgut volvulus, trisomy 21, fetal autopsy, whirlpool appearance, ultrasound, amniocentesis

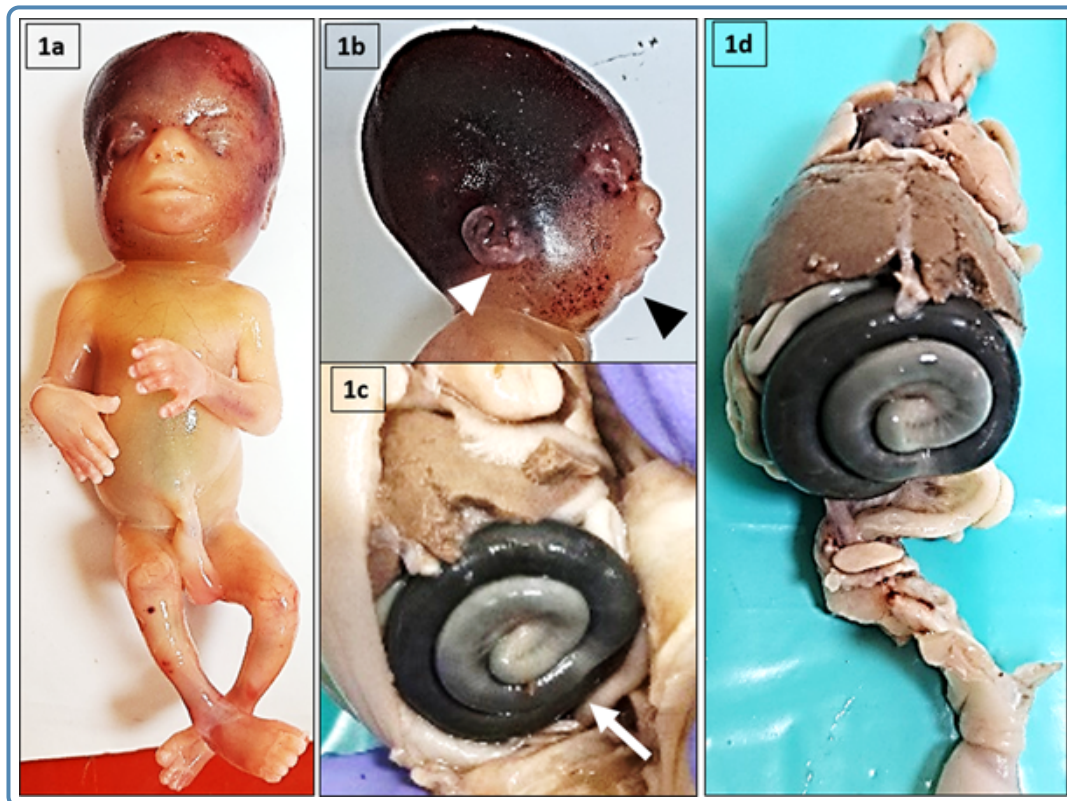
## Introduction

Down syndrome is the most common numerical chromosomal abnormality with an incidence of 1 in 850 live births and is characterized by intellectual disability, dysmorphism and congenital malformations (Al-Nbaheen et al., 2018). Anomalies of the gastrointestinal tract make up 5-7% of all congenital malformations and these are due to atresia or stenosis (Morris et al., 2014). In the present case report, we describe a rare case of midgut volvulus without atresia which was incidentally discovered in a fetus with trisomy 21.

## Patient Details

A 29-year-old second gravida (G2A1) was referred at 19 weeks 1 day (19 weeks 6 days as per the first trimester dating scan done at 6 weeks 1 day) of gestation in view of ventriculomegaly and short long bones in the fetus. The couple were married non consanguineously, and the pregnancy was conceived spontaneously after one previous spontaneous abortion in the first trimester. In the present pregnancy, the first trimester was uneventful. Aneuploidy screening was not done. On ultrasonographic evaluation, diameters of atria of both lateral ventricles of the brain were more than 15 mm, suggestive of severe ventriculomegaly along with hypoplastic cerebellum (cerebellar diameter - 17.2 mm; -2.2 SD for 19 weeks of gestation). The bowel was slightly echogenic. Growth parameters and measurements of all long bones fell below -3 SD (corresponding to 16 weeks of gestation) suggestive of potential growth restriction in the fetus. No other soft marker or malformation was noted. A chromosomal etiology was suspected, and the couple opted for amniocentesis for fetal karyotype, after counselling. In view of severity of the anomalies and poor prognosis for growth and survival, the couple decided to terminate the pregnancy and submitted the fetus for autopsy.

On external examination, the fetus was female with subcutaneous edema below the chin and lower jaw extending to the rest of the body. Slight retrognathia and low set ears were noted in the face with intact lips and palate (**Figure 1a and b**). Both upper and lower limbs were devoid of any abnormality with no ano-rectal malformation. The cord had three vessels. The anthropometric measurements and fetal weight fell below the 5<sup>th</sup> centile for gestational age of 19 weeks and were indicative of growth restriction in the fetus. Internal examination of the brain confirmed the presence of ventriculomegaly and hypoplastic



**Figure 1** Images of fetal autopsy. **1a:** Front profile of the fetus. **1b:** Side profile of the face with white pointer towards the low set ears and the black pointer showing the retrognathia. **1c:** White arrow pointing towards the characteristic whirlpool coiling of the midgut in situ. **1d:** Internal examination of the fetal organs with the midgut volvulus.

cerebellum. On opening the abdomen, loops of dilated gut were coiled in a characteristic whirlpool pattern suggestive of volvulus (**Figure 1c and d**). No evidence of atresia or stenosis were found on examination of the midgut. No malformation was noted in the esophagus, trachea, lungs, and heart with an intact diaphragm. Remainder of the abdomen and pelvic organs were unremarkable with normal histopathology. Fetal karyotype revealed free trisomy 21.

## Discussion

Association of midgut volvulus with trisomy 21 is extremely rare. Midgut volvulus is seen in 3.9/10,000 live births and can be picked up antenatally in the third trimester by various ultrasound findings of which dilated stomach and bowel, polyhydramnios and whirlpool sign are the most frequent (Schulman et al., 1993; Shen et al., 2022). Review of videos of prenatal ultrasonography done

in our case did not reveal anything other than echogenic bowel around the umbilicus suggesting that at early gestation the whirlpool sign may not be obvious on ultrasonography. Echogenic bowel has been reported in 11.4% cases at a median gestational age of 31 weeks (Shen et al., 2022).

The most common cause of midgut volvulus is intestinal malrotation. Other rare associations include intestinal atresia and duplication, cystic fibrosis, and segmental mesenteric defect (Shen et al., 2022). Only a single case series with 11 cases reported one fetus with trisomy 21 (Yang et al., 2022).

This is the second reported case of midgut volvulus seen with trisomy 21. Such findings can be missed in the second trimester anomaly scan. In our case it was detected incidentally at fetal autopsy, reiterating the importance of doing a systematic fetal autopsy in uncovering rare developmental anomalies.

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**Conflict of interest:** None

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