

# Obstetric Ultrasound Challenge

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## ABSTRACT

Obstetric ultrasound is the primary modality for detecting fetal anomalies that affect antepartum care of the mother and care of the neonate after delivery. The following cases represent the challenging cases imaged in our Fetal Diagnostic Center.

**Objectives:** Determine the fetal anomaly. Understand the work-up for the associated fetal anomaly.

**Keywords:** Fetal anomaly, Hemoglobin bart, Situs inversus totalis, Trisomy 18, Ultrasound challenge.

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## INTRODUCTION

Obstetric ultrasound is the primary modality for detecting fetal anomalies that affect antepartum care of the mother and care of the neonate after delivery. The following cases represent the challenging cases imaged in our Fetal Diagnostic Center.<sup>1,2</sup>

## CASE REPORTS

### Case 1 (Figs 1A to D)

Ms MD was a 20-year-old primiparous patient who presented at 20 weeks 6 days with early onset intrauterine growth restriction (IUGR).

Ultrasound findings included:

- Early IUGR
- Echogenic bowel
- Cardiomegaly with hyperdynamic state and prominent vasculature
- Kyphosis
- Abnormal nuchal fold

The most likely diagnosis was:

- Hemoglobin barts.

## Clinical Course

Ms MD underwent an amniocentesis that returned with inability to perform a karyotype, elevated alpha-fetal protein (AFP) at 3.03 MoM, negative anticholinesterase, and negative infectious disease work-up. The maternal work-up returned positive for alpha-thalassemia. Further ultrasound findings were worsening oligohydramnios, fetal hydrops, and abnormal umbilical and middle cerebral Doppler. Delivery was recommended, but the patient declined. She was followed with weekly blood pressure, urine dips and monitoring for signs and symptoms of pre-eclampsia. Ms MD was delivered at 28 weeks due to preterm labor (PTL) and Mirror syndrome.<sup>6</sup>

## Discussion

Alpha thalassemia is a form of hemoglobinopathy with the most severe form in hemoglobin barts. Its inheritance pattern is autosomal recessive. Found most commonly in patients from Asia or Eastern Mediterranean, this occurs when all four alpha-globin chains are deleted. It affects the fetus due to its ability to steal oxygen from the fetus, which usually dies prior to birth or shortly thereafter due to fetal hydrops. Diagnosis is made through genetic testing as hemoglobin electrophoresis alone does not detect alpha-thalassemia. It, however, can be an initial indicator.<sup>6</sup>

### Case 2 (Figs 2A to D)

Ms SD was a G3P0020 at 32 weeks who presented with concordant dichorionic-diamniotic twins. She had a sequential screen and nuchal translucency (NT) on both twins performed. The NT for each twin was normal at 11 weeks. The sequential screen returned with the following abnormalities<sup>3</sup>:

- AFP 2.91 MoM
- Estriol 1.02 MoM
- hCG 8.49 MoM
- Inhibin 10.48 MoM
- Down syndrome age 1:842
- Down syndrome age + analytes 1:171.

An ultrasound was performed at 18 weeks 5 days with normal anatomy noted in both twins. The amniocentesis returned normal with the following results:

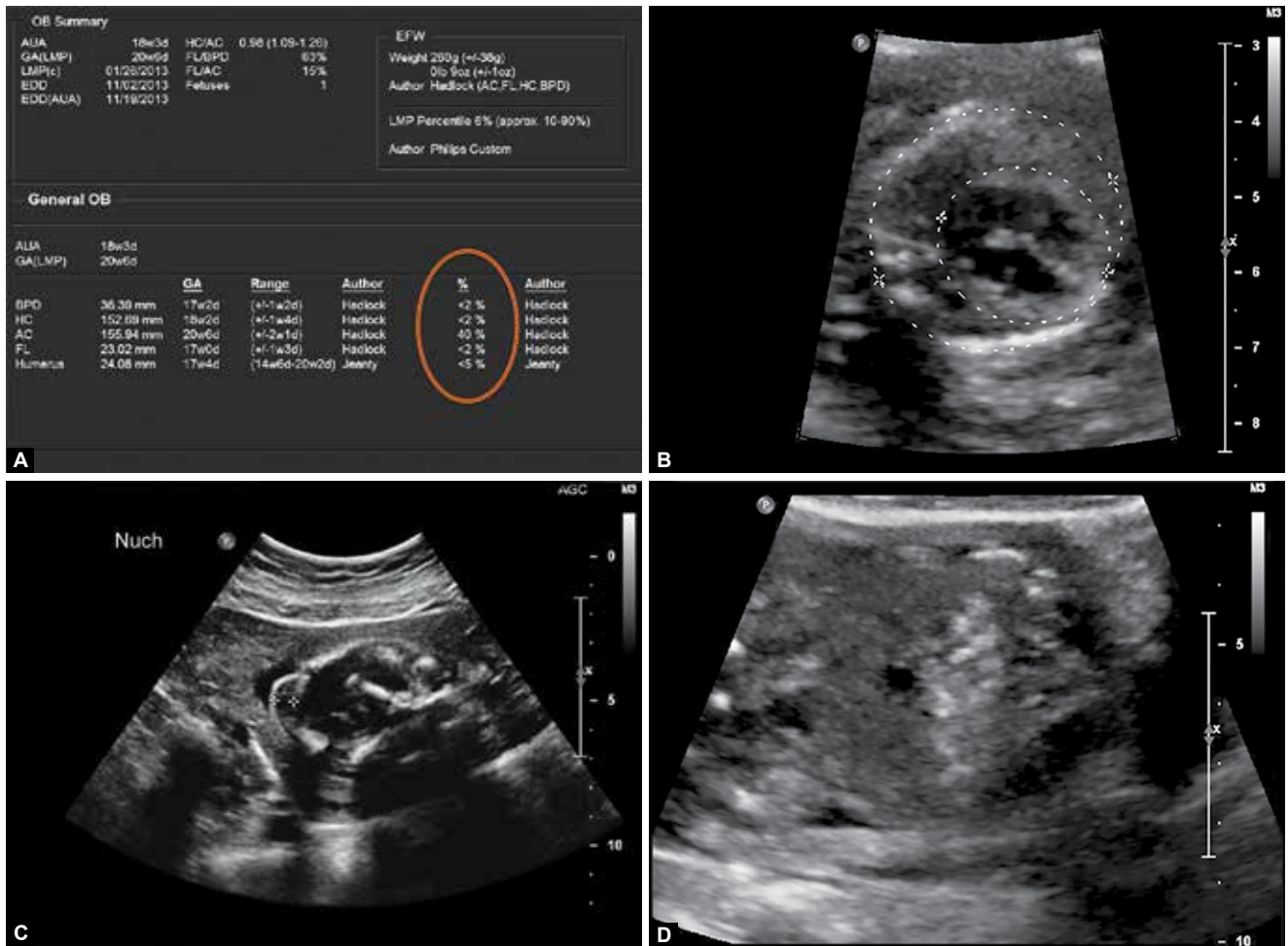
- A: 46,XX, AFP 0.58 MoM
- B: 46,XY, AFP 0.59 MoM

A fetal echo on the twins was performed at 29 weeks.

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**Figs 1A to D:** (A) Biometry demonstrating fetal growth restriction, (B) fetal cardiomegaly, (C) abnormal nuchal fold and (D) echogenic bowel (Source: Kapiolani medical center for women and children’s fetal diagnostic center)

The most likely diagnosis for Twin B is (Figs 2A to D):

- Situs inversus totalis
- Ventricular septal defect (VSD).

**Clinical Course**

The fetal echo for twin A was normal. Twin B was found to have situs inversus totalis, complete dextrocardia with anatomically connections and a small, muscular VSD near the heart apex. The patient was followed monthly for interval growth, antepartum testing begun at 34 weeks and delivered at 35 weeks via cesarean delivery for preterm premature rupture of membranes. Twin B was evaluated postnatally by the pediatric cardiologist.

**Discussion**

Dextrocardia is a congenital heart defect where the apex of the heart is pointed toward the right instead of the left. It can be associated with transposition of the great vessels or have correct connections as in Twin B above. Situs inversus totalis is when the organs are also located anatomically on the opposite side. If all the connections are anatomically correct but just inverted, then the fetus is relatively unaffected.<sup>4,5</sup>

**Case 3 (Figs 3A to D)**

Ms LM was a 39-year-old primiparous patient who presented to the Fetal Diagnostic Center at 20 weeks 1 day for a fetal anatomical survey and unsure last menstrual period (LMP). Ms LM was suspected to have a skeletal dysplasia with a height of 4 feet 2 inches.

Ultrasound findings included polyhydramnios, lemon sign with hypoplastic cerebellum and absent cavum septum pellucidum (CSP), micrognathia, omphalocele and clubbed feet and hands.

**What is the most likely Chromosome Abnormality?**

- Trisomy 18

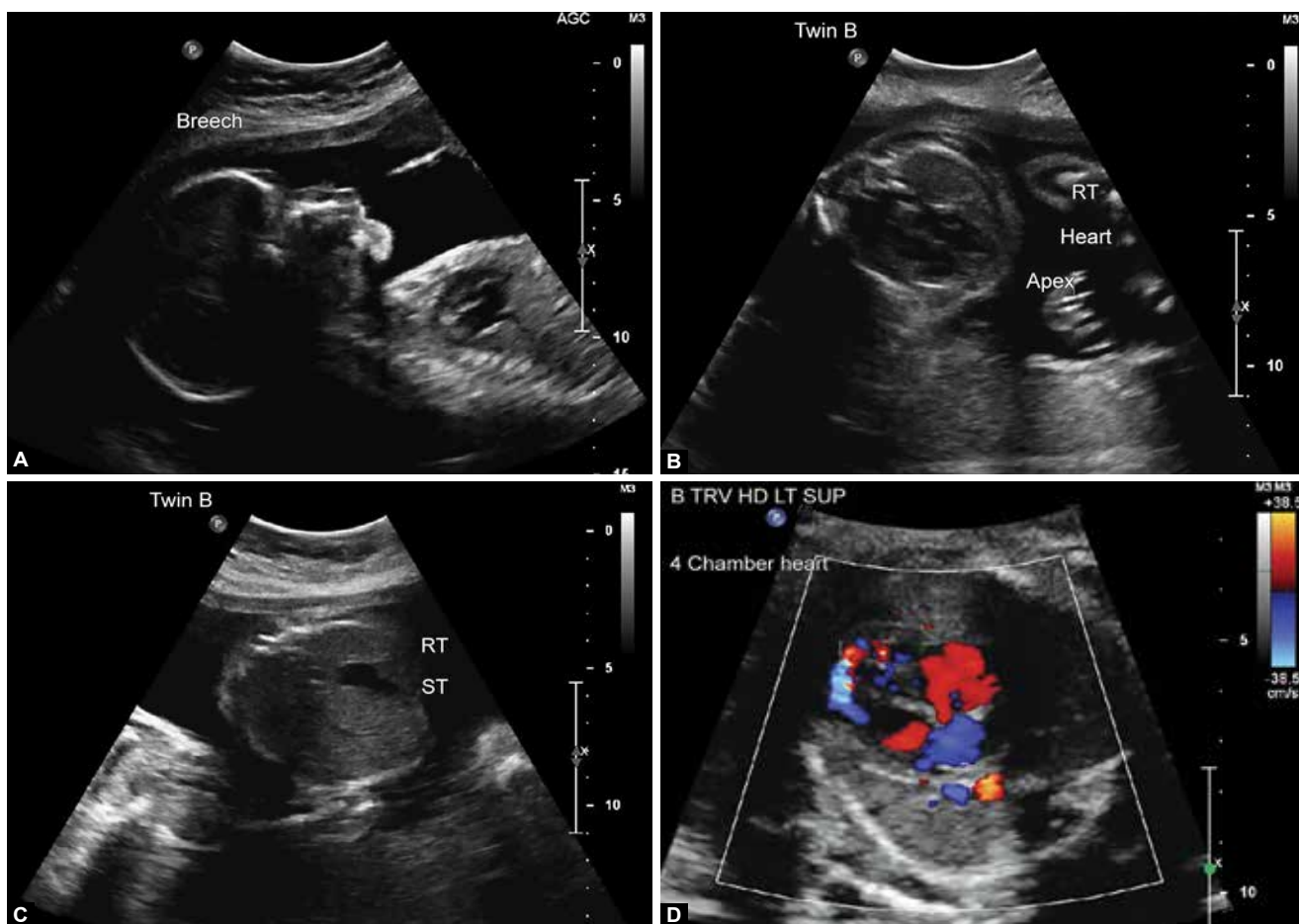
**Clinical Course**

The patient had an amniocentesis that showed Trisomy 18. She opted for termination at 20 weeks 1 day by induction of labor with misoprostol.

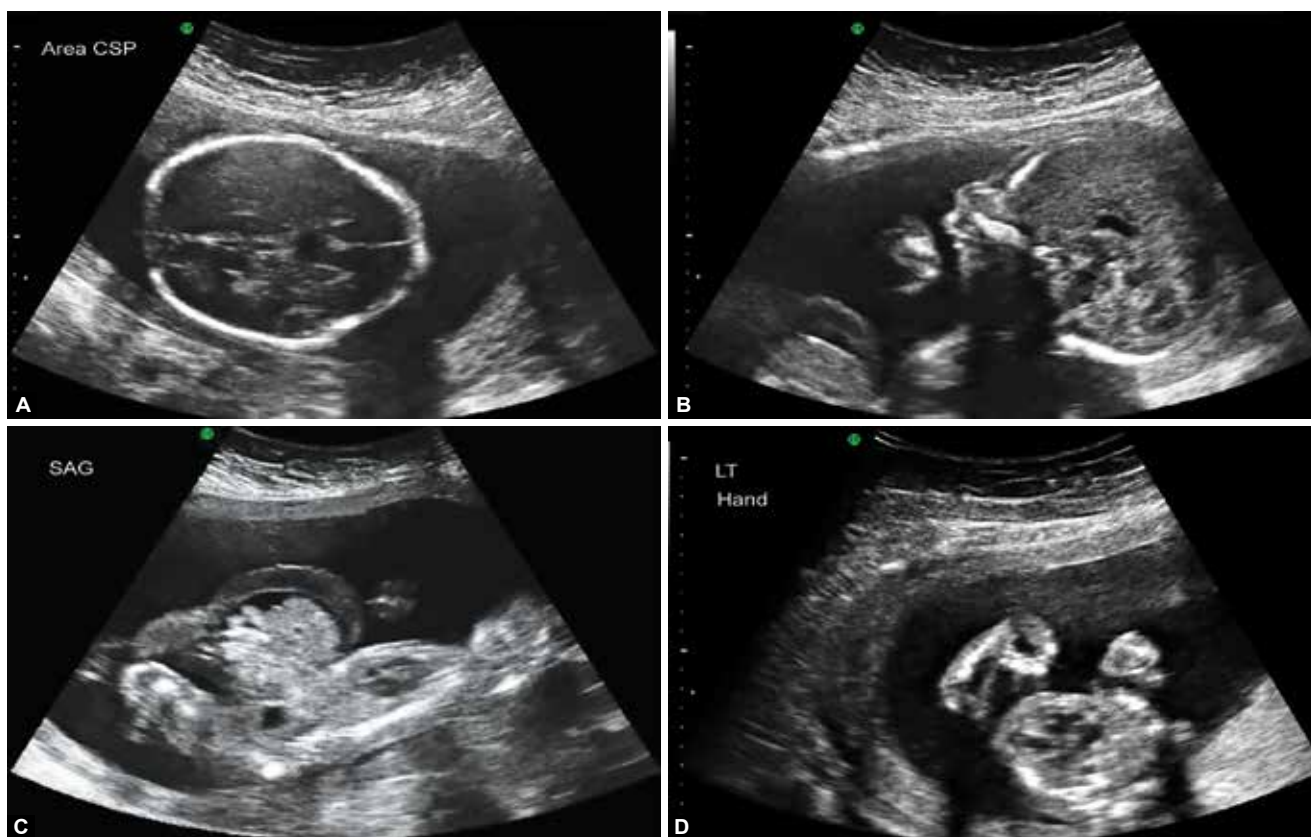
**Discussion**

Trisomy 18 is one of the three chromosomal defects associated with aneuploidy that is routinely screened for





**Figs 2A to D:** (A) Establishing situs, (B) demonstrating situs, (C) showing stomach is on right and (D) shows VSD  
 (Source: Kapiolani medical center for women and children's fetal diagnostic center)



**Figs 3A to D:** (A) Absent CSP with lemon sign, (B) Micrognathia, (C) Omphalocele and (D) Clubbed extremity  
 (Source: Kapiolani medical center for women and children's fetal diagnostic center)

in obstetrics. The others are Trisomy 21 and Trisomy 13.<sup>3</sup> Also known as Edward's syndrome, it is associated with rocker-bottom feet, clenched hands, omphalocele, micrognathia and congenital heart defects on prenatal ultrasound. It is diagnosed by genetic amniocentesis. Most fetus die intrauterine but if survive after delivery, then die within a few days. It can be diagnosed earlier with chorionic villus sampling (CVS).

## REFERENCES

1. ISUOG Education Committee recommendations for basic training in obstetric and gynecological ultrasound. *Ultrasound Obstet Gynecol* 2014;43:113-116.
2. American Institute of Ultrasound in M. AIUM Practice Guideline for the performance of an antepartum obstetric ultrasound examination. *J Ultrasound Med* 2003;22:1116-1125.
3. Bestwick JP, Wald NJ. Sequential integrated antenatal screening for Down's syndrome, trisomy 18 and trisomy 13. *J Med Screening* 2015.
4. Copel JA. Obstetric ultrasound terminology. *Seminars in Perinatol* 2013;37:305-306.
5. Oztunc F, Besikci R, Turkoglu H, Saltik IL. Prenatal echocardiographic diagnosis of situs inversus totalis and transposition of the great arteries: a case report. *Turkish J Pediatr* 2001;43:345-347.
6. Winichagoon P, Sithongdee S, Kanokpongsakdi S, Tantisirin P, Bernini LF, Fucharoen S. Noninvasive prenatal diagnosis for hemoglobin Bart's hydrops fetalis. *Int J Hematol* 2005;81:396-399.