



First Trimester Diagnosis of Congenital Heart Disease

Nikhil Gholkar^{1,2} · Chanchal Singh¹ · Anita Kaul¹ · Kanwal K. Kapur³

Received: 18 May 2015 / Accepted: 27 July 2015 / Published online: 2 September 2015
© Society of Fetal Medicine 2015

A 26-year-old primigravida came to our Fetal Medicine Center for first trimester aneuploidy screening at 13 weeks. Ultrasound was done using Voluson E8 (GE Healthcare®, Milwaukee, WI, USA) with convex abdominal 4–8 MHz transducer and transvaginal 15–20 MHz transducer. On abdominal imaging, there was cardiomegaly with tricuspid regurgitation. On transvaginal scan, clear cardiac disproportion with a small left atrium and ventricle were seen (Fig. 1). On application of color Doppler, the flow across the mitral and aortic valves was negligible with hyperdynamic flow on the right side with tricuspid regurgitation (Figs. 2, 3). Foramen ovale was opening on the right side and the flow in the arch of aorta was reversed.

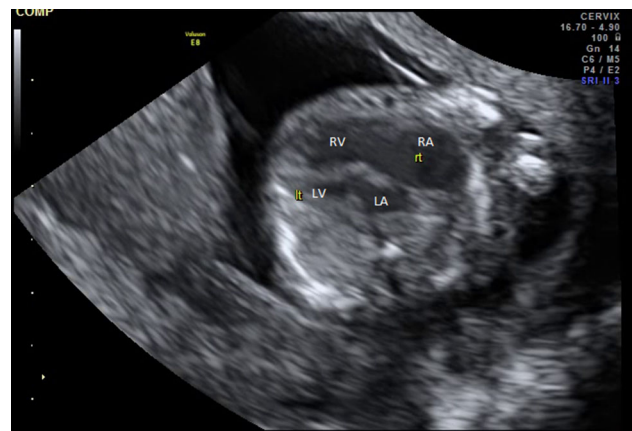


Fig. 1 An axial image of the fetal chest on transvaginal sonography at 13 weeks showing a grossly-enlarged heart with a clear ventricular disproportion (*Right>>>Left*)

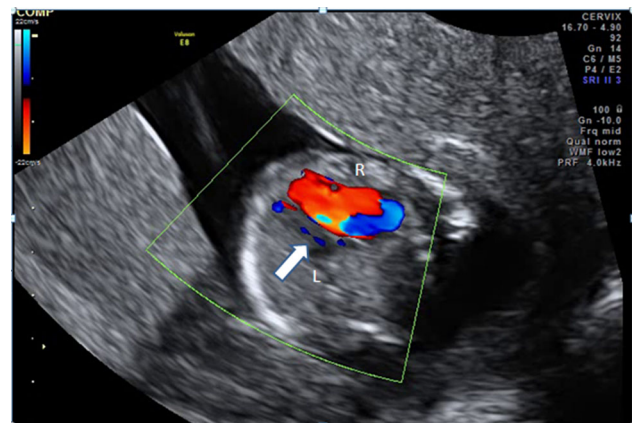


Fig. 2 On application of *color Doppler* on a transvaginal axial image of the fetal heart, negligible flow is seen across the mitral valve while hyperdynamic flow is seen across the tricuspid valve

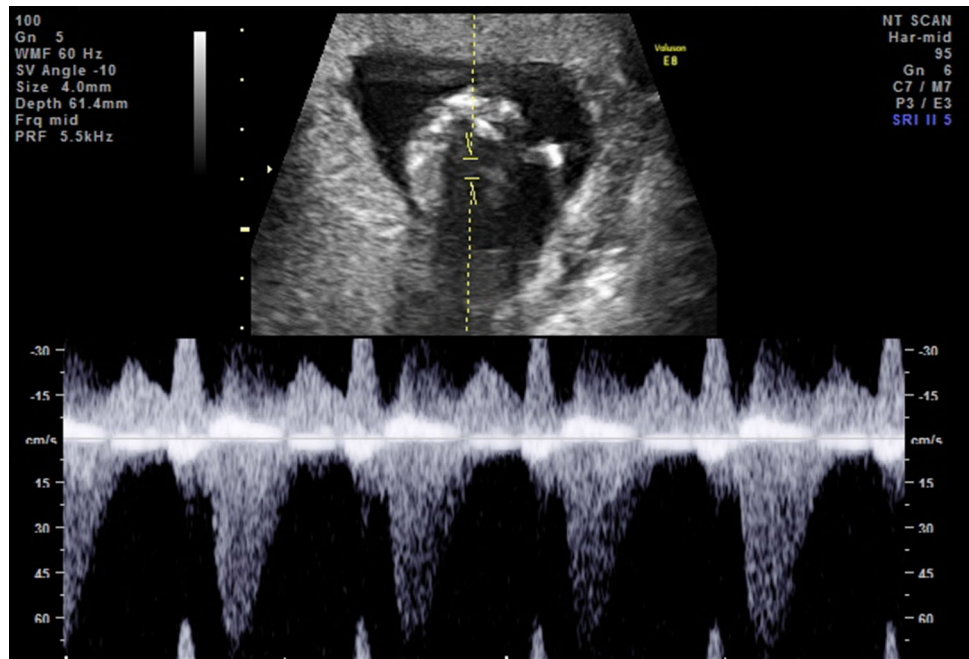
✉ Nikhil Gholkar
drnikhilgholkar@gmail.com

¹ Apollo Centre for Fetal Medicine, Indraprastha Apollo Hospitals, New Delhi, India

² B 704, Parkland 1, Raheja Estate, Borivali East, Mumbai 400066, India

³ Department of Cardiology, Indraprastha Apollo Hospitals, New Delhi, India

Fig. 3 Transabdominal ultrasound demonstrating of regurgitation across the tricuspid valve suggestive of hyperdynamic circulation on the right side



All the above features confirmed the diagnosis of hypoplastic left heart syndrome (HLHS). HLHS constitutes a whole spectrum of cardiac abnormalities involving under development of the left side of the fetal heart and thus, leading to inadequate systemic circulation [1]. HLHS has an incidence of about 1–2/10,000 live births but can account for up to 9 % of all cases of congenital heart disease [2]. On initial transabdominal cardiac screening, the alarming signs can be an abnormal-looking four-chamber view, cardiomegaly, or tricuspid regurgitation which should trigger a detailed transvaginal evaluation of the fetal heart. The most common condition to differentiate from HLHS is coarctation of aorta and the most difficult condition to differentiate is critical aortic stenosis. The visualization of the flow across the mitral and aortic valves and also across the ascending and aortic arch is the key to final diagnosis [3]. The presence of tricuspid regurgitation and/or restriction of flow across the foramen ovale are features associated with poor prognosis [1, 3]. There is a high incidence of chromosomal abnormalities (4 %–5 %) and also of other structural defects (10 %–25 %), therefore, a detailed evaluation of the other anatomy by ultrasound and offering a diagnostic test for chromosomal abnormalities should be considered. [3–5]. Although, the prognosis for these fetuses with surgical correction has improved over the years, the overall outlook still remains poor [6].

After a detailed discussion on possible management options and long-term prognosis, the couple opted for termination of pregnancy. The karyotype of the fetus was

normal. This case highlights the significance of an early fetal echocardiography after an initial abnormal cardiac screening. Early diagnosis can be made using high-frequency transabdominal and transvaginal sonography as early as 12–13 weeks. This helps in early decision making, safe termination if opted for, and minimizing emotional trauma.

Compliance with Ethical Standards

Conflict of Interest None.

References

1. Rychik J. Hypoplastic left heart syndrome: from in utero diagnosis to school age. *Semin Fetal Neonatal Med.* 2005;10:553–66.
2. Hoffman JI, Kaplan S. The incidence of congenital heart disease. *J Am Coll Cardiol.* 2002;39:1890–900.
3. Abuhamad A, Chaoui R. *A practical guide to fetal echocardiography: normal and abnormal hearts.* 2nd ed. Philadelphia: Lippincott Williams & Wilkins; 2010.
4. Raymond FL, Simpson JM, Sharland GK, et al. Fetal echocardiography as a predictor of chromosomal abnormality. *Lancet.* 1997;350:930.
5. Natowicz M, Chatten J, Clancy R, et al. Genetic disorders and major extracardiac anomalies associated with the hypoplastic left heart syndrome. *Pediatrics.* 1988;82:698–706.
6. Galindo A, Nieto O, Villagra S, et al. Hypoplastic left heart syndrome diagnosed in fetal life: associated findings, pregnancy outcome and results of palliative surgery. *Ultrasound Obstet Gynecol.* 2009;33:560–6.