EC Paediatrics Special Issue - 2017

On which One Newborn Babies should Echocardiography be Performed?

Case Report

Dr. Hashem E Khosroshahi*

Department of Pediatrics, Pediatric Cardiology Unit, Bozok University Medical Faculty, Yozgat, Turkey

***Corresponding Author:** Dr. Hashem E Khosroshahi, Department of Pediatrics, Pediatric Cardiology Unit, Bozok University Medical Faculty, Yozgat, Turkey.

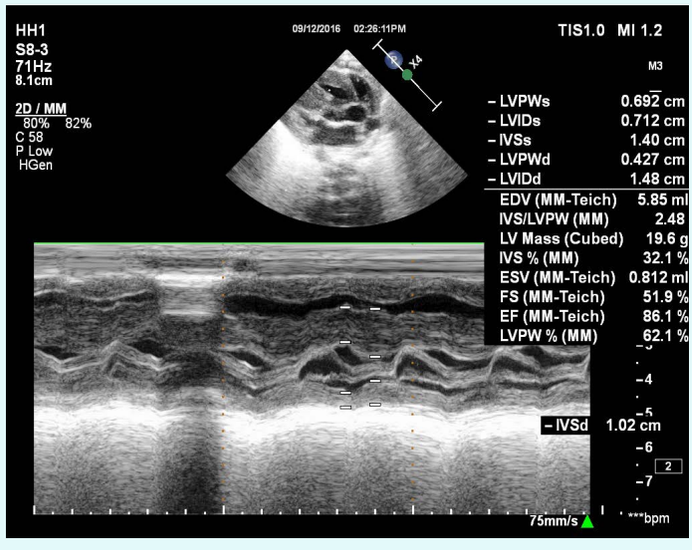
Received: November 14, 2017; **Published:** November 22, 2017

Introduction

Hypertrophic cardiomyopathy (HCM), obstructive or non-obstructive, may be diagnosed among newborn babies without any related sign and symptoms, and can raise the question if a transthoracic echocardiographic (TTE) examination should be carried out for all newborn babies who look apparently normal? This may remain as a dilemma among neonatologist, pediatricians and pediatric cardiologists during their routine daily practice.

Case Report

A 3970g (height 86 percentile, weight 95 percentile) male newborn was delivered via cesarean section at 36 weeks' gestation, of healthy, nonconsanguineous parents, with history of gestational diabetes and preeclampsia. His mother (gravida 2, para 2) was 34-year-old and his father was 37-year-old at the time of the delivery. The Apgar scores at 1 and 5 minutes after birth were 6 and 9, respectively. The initial vital signs were body temperature 36.5oC, heart rate 130/min and respiratory rate 55/min. A short 1/6 systolic murmur was heard on upper left sternal border. Physical findings were otherwise normal. Transthoracic echocardiography revealed severe septal hypertrophy, mild coarctation of aorta (Figure 1A, B, C) and mild mitral prolapse. Family history was negative for hypertrophic cardiomegaly. Patient has been followed up closely and the septal hypertrophy was decreased in 4 months dramatically without any treatment (Figure 1D).



Citation: Dr. Hashem E Khosroshahi. "On which One Newborn Babies should Echocardiography be Performed?". EC Paediatrics SI.01 (2017): 16-20.

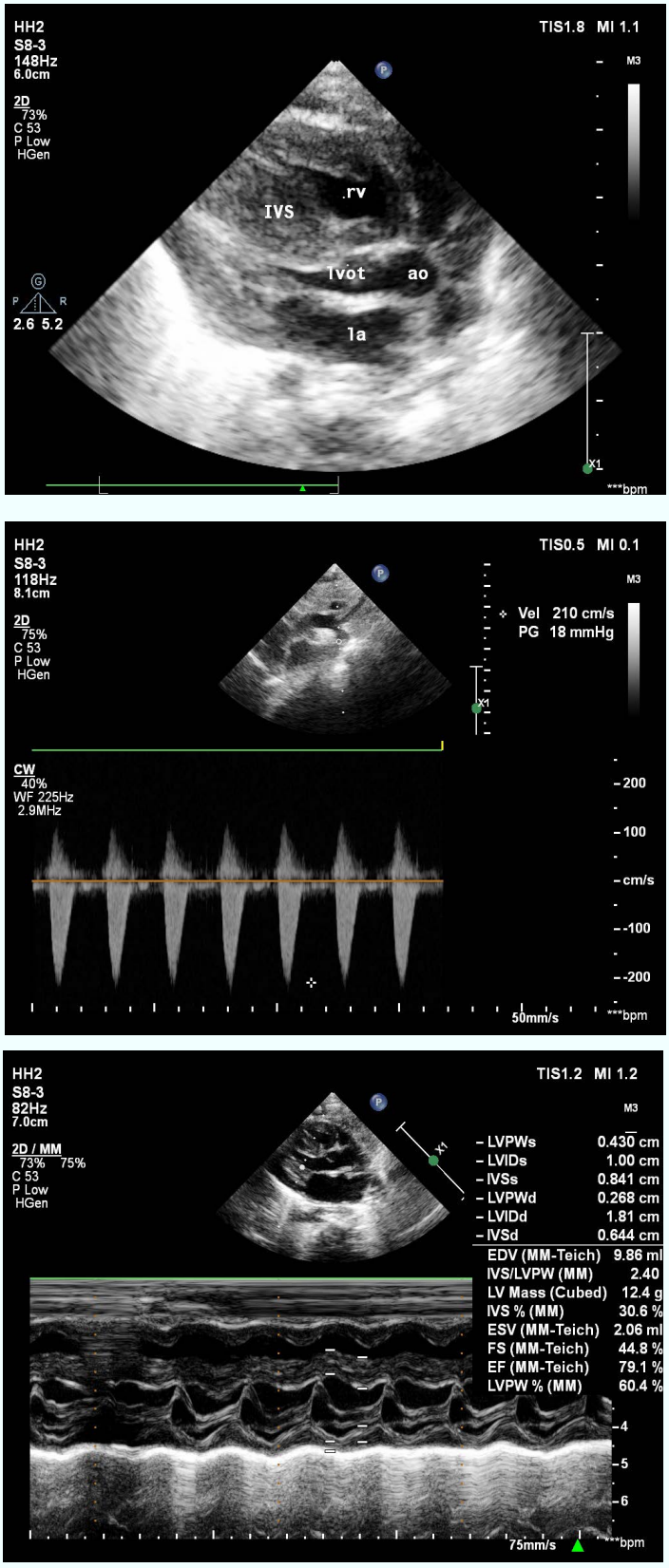


Figure 1: *A: M-mode, B: 2-D and C: Doppler echocardiography of the patient showing significant septal hypertrophy and mild coarctation of aorta at first day of life. Septal hypertrophy was diminished dramatically after 4 months without any treatment (D).*

Patients and Findings

Retrospective chart review of all patients age 1 day-16 years managed by pediatric cardiology outpatient department of our institution over the last 13 months (September 2016-November 2017) period revealed that 134/3014 (4,4%, M/F = 86/48) patients had echocardiographically determined septal hypertrophy. Fifty (1,7%, M/F = 30/20) patients with HCM, were the babies under 40 days of age, of whom 9 (18%) patients were children of mothers with known gestational diabetes. Signs and symptoms of the neonates with HCM are summarized in table 1. As shown in table 2 we found different congenital heart defects co-existing with HCM.

Signs and symptoms	N = 50
Murmur	33
History of gestational diabetes	9
Tachypnea/Respiratory difficulty	3
Cleft lip	1
Down syndrome	1
History of CHD	1
Prematurity	1
Tachycardia	1

Table 1: *Signs and symptoms of patients with hypertrophic cardiomyopathy under 40 days of age.*

Cardiac lesions	N = 50
Atrial septal defect	21
Patent foramen ovale	15
Patent ductus arteriosus	12
Main and peripheral Pulmonary stenosis	9
Ventricular septal defect	8
Persistent pulmonary hypertension	6
Aortic stenosis	3
Coarctation of aorta	2
Endocardial fibroelastosis	1
Bicuspid Aorta	1
Mitral insufficiency	1
Without any lesion	3

Table 2: Cardiac lesions co-exist in children with hypertrophic cardiomyopathy under 40 days of age.

Discussion

The prevalence of HCM in the general population is about 0.2% (1:500) [1]. HCM is common in newborns of mothers with gestational diabetes mellitus and in neonates with congenital hyperinsulinism [2-4].

HCM have been described in infants suffer from valvular and supravalvular aortic stenosis, aortic hypoplasia, bicuspid aortic valve, aortic coarctation, mitral valve prolapse, valvular and supravalvular pulmonary artery stenosis and septal defects [5,6]. The typical cardiac defect of supravalvular aortic stenosis is seen in around 70% of patients with HCM [7]. Intramural (small vessel) coronary artery disease has been described in young children with HCM [8].

There are cases were reported as primary or idiopathic HCM which were diagnosed clinically by the presence of left ventricular hypertrophy (often asymmetrical) in the absence of congenital heart disease, hypertension, valve lesions, infection or other systemic illnesses [9]. It is known that the HCM is the second commonest form of heart muscle disease affecting children and adolescents and is a leading cause of sudden death in young athletes and is one of the most common monogenic cardiovascular disorders and is caused by mutations in a variety of genes encoding proteins of the cardiac sarcomere [9-12]. Genetic testing is becoming increasingly incorporated into clinical practice in the assessment of cardiomyopathies. Recent reviews highlight the genetic basis and pathogenesis of HCM [13-16].

One of our patients presented with cleft lip (1/134) without any cardiac lesion. Hypertrophic cardiomyopathy co-existing with cleft lip/palate may be seen as part of Vici syndrome, Cornelia de Lange syndrome, Noonan syndrome, Costello syndrome and some other syndromes [17,18].

Conclusion

Years ago Maron., *et al*. stated that a single echocardiographic examination of young relatives of patients with hypertrophic cardiomyopathy may not exclude this disease [19]. We strongly recommend that the routine TTE and EKG for all newborn infants with or without congenital heart defect and any syndromic disease, and particularly in the setting of a so-called innocent murmur or respiratory difficulties should be performed.

Bibliography

1. Maron BJ. "Hypertrophic Cardiomyopathy: A Systematic Review". *Journal of the American Medical Association* 287.10 (2002): 1308-1320.
2. Way GL., *et al*. "The natural history of hypertrophic cardiomyopathy in infants of diabetic mothers". *Journal of Pediatrics* 95.6 (1979): 1020-1025.
3. Breitweser JA., *et al*. "Cardiac septal hypertrophy in hyperinsulinemic infants". *Journal of Pediatrics* 96 (1980): 535-539.
4. Huang TT., *et al*. "Hypertrophic cardiomyopathy in neonates with congenital hyperinsulinism". *Archives of Disease in Childhood - Fetal and Neonatal Edition* 98.4 (2013): F351-F354.
5. Somerville J and Becú L. "Congenital heart disease associated with hypertrophic cardiomyopathy". *British Heart Journal* 40.9 (1978): 1034-1039.
6. Abu-Sulaiman RM and Subaih B. "Congenital heart disease in infants of diabetic mothers: echocardiographic study". *Pediatric Cardiology* 25.2 (2004): 137-140.
7. Moser-Bracher A., *et al*. "Severe neonatal hypertrophic obstructive cardiomyopathy". *Swiss Society of Neonatology* (2003).
8. Maron BJ., *et al*. "Intramural ("small vessel") coronary artery disease in hypertrophic cardiomyopathy". *Journal of the American College of Cardiology* 8.3 (1986): 545-557.
9. Moak JP and Kaski JP. "Hypertrophic cardiomyopathy in children". *Heart* 98.14 (2012): 1044-1054.
10. Lekanne Deprez RH., *et al*. "Two cases of severe neonatal hypertrophic cardiomyopathy caused by compound heterozygous mutations in the MYBPC3 gene". *Journal of Medical Genetics* 43.10 (2006): 829-832.
11. Cirino AL and Ho C. "Hypertrophic Cardiomyopathy Overview". *Gene Reviews Advanced Search* (2014).
12. Cox GF., *et al*. "Factors associated with establishing a causal diagnosis for children with cardiomyopathy". *Pediatrics* 118.4 (2006): 1519-1531.

13. Ware SM. "Genetic diagnosis in pediatric cardiomyopathy: clinical application and research perspectives". *Progress in Pediatric Cardiology* 31.2 (2011): 99-102.

14. McBride KL and Garg V. "Impact of Mendelian inheritance in cardiovascular disease". *Annals of the New York Academy of Sciences* 1214 (2010): 122-137.

15. Alcalai R., *et al*. "Genetic Basis of Hypertrophic Cardiomyopathy: From Bench to the Clinics". *Journal of Cardiovascular Electrophysiology* 19.1 (2008): 104-110.

16. Baertling F., *et al*. "Mutations in COA6 cause Cytochrome c Oxidase Deficiency and Neonatal Hypertrophic Cardiomyopathy". *Human Mutation* 36.1 (2015): 34-38.

17. Byrne S., *et al*. "Vici syndrome: a review". *Orphanet Journal of Rare Diseases* 11 (2016): 21.

18. Wenger TL., *et al*. "Novel findings of left ventricular non-compaction cardiomyopathy, microform cleft lip and poor vision in patient with SMC1A -associated Cornelia de Lange syndrome". *American Journal of Medical Genetics Part A* 173.2 (2017): 414-420.

19. Maron BJ., *et al*. "Development and progression of left ventricular hypertrophy in children with hypertrophic cardiomyopathy". *New England Journal of Medicine* 315.10 (1986): 610-614.