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Echocardiographic Diagnosis of Heart Disease in Apparently Healthy Adolescents

Julia Steinberger, MD; James H. Moller, MD; James M. Berry, RDMS; and Alan R. Sinaiko, MD

ABSTRACT. Objective. Comprehensive data are currently unavailable on the prevalence of cardiac abnormalities in children after the newborn/infant period. The present report describes the prevalence of echocardiographically detected cardiac disease in a cohort of randomly selected healthy junior high school children.

Methods. The cohort for this report consists of 357 children (mean age: 13 years) randomly selected after blood pressure screening of 12,043 fifth through eighth grade students and having an echocardiographic examination as part of a study of insulin resistance in childhood.

Results. A physical examination performed by a board-certified pediatrician reported no cardiac abnormalities. However, echocardiography and Doppler studies identified 13 (3.6%) children (7 males and 6 females), with previously unknown cardiac abnormalities, as follows: abnormal mitral valve with mitral regurgitation (4), bicuspid aortic valve (2), atrial septal defect (2), coronary artery to pulmonary artery fistula (1), patent ductus arteriosus (1), pulmonary hypertension (1), cardiomyopathy (1), and pulmonary artery stenosis (1). Physical examination performed by a pediatric cardiologist detected abnormal cardiac findings in 7 (54%) of the children. Cardiac catheterization was required in 3 for additional diagnostic evaluation and in 2 for therapeutic intervention; 1 patient underwent open-heart surgery. Bacterial endocarditis prophylaxis was recommended in 8 (62%) of the 13 children.

Conclusions. The results suggest that: 1) clinically significant cardiac disease in childhood is more prevalent than previously reported; and 2) improved screening methods should be considered to detect asymptomatic but significant cardiac abnormalities that may result in long-term complications. Pediatrics 2000;105:815–818; echocardiography, prevalence, incidence, heart disease, children.

The prevalence of heart disease in children and adolescents has not been determined. Although studies in infants have shown a prevalence of congenital heart disease of ~2 to 8 cases per 1000 live births in the United States,1 and it has been estimated that the worldwide prevalence of congenital heart disease is ~4 to 12 per 1000 births,2,3 similar population studies have not been performed in older children.

Early detection of cardiac disease in children of all ages is important to prevent serious complications of what would otherwise be mild, and potentially easily corrected, disease and to institute prophylaxis of bacterial endocarditis. The present report describes the prevalence of echocardiographically detected cardiac disease in a cohort of randomly selected healthy junior high school children. The results suggest that there are a significant number of children with undetected cardiac disease and document the limitations of physical examination, even by trained pediatric cardiologists, in making the diagnosis.

METHODS

The study population was participating in a longitudinal evaluation of insulin resistance in childhood. Their mean age was 13 years (range: 11–15 years). After blood pressure screening of 12,043 fifth through eighth grade Minneapolis Public School students (93% of all eligible students), the children were randomly selected for participation, with stratification according to sex, race, and blood pressure (50% from the upper 25 percentiles and 50% from the lower 75 percentiles). A total of 401 students were recruited and underwent a complete physical examination by board-certified pediatricians. Of these, 357 students (193 male and 164 female; 77 black and 280 white) had an insulin clamp procedure performed at the University of Minnesota Clinical Research Center. This group forms the cohort for the present report. Complete 2-dimensional echocardiograms and Doppler studies were obtained on all participants in the Pediatric Echocardiography Laboratory at the University of Minnesota, using Hewlett-Packard echocardiographic equipment. All studies were performed by the same technician and were read by the same pediatric cardiologist, who was blinded to the physical findings of the subjects. All subjects found to have an abnormal echocardiogram were referred for additional evaluation and treatment by the pediatric cardiologist who read the echocardiogram. Isolated asymptomatic patent foramen ovale was found in 3 (8%) of the children, but they are not included as cardiac abnormalities in this report.

RESULTS

Cardiac abnormalities were found in 13 of the 357 children, based on 2-dimensional echocardiogram and Doppler studies. The clinical data on these children are described in Table 1. There were no significant differences regarding age, sex, race, height, sexual maturity, systolic and diastolic blood pressure, and heart rate among the 13 children with cardiac abnormalities and the remaining 344 children without cardiac abnormalities, but children with cardiac abnormalities had lower weight and body mass index. No significant clinical or cardiac differences were found between blacks (n = 3) and whites (n = 10) or males (n = 7) and females (n = 6).

None of the 13 children had abnormal physical findings on an examination performed by a board-certified pediatrician. As noted in Table 2, only 7 of...
the 13 (54%) had abnormal cardiovascular findings on a physical examination performed by a pediatric cardiologist who at the time of the examination was aware of the echocardiographic results. Catheterization and/or surgery were performed in 6 children, and 7 did not require any additional procedures (Table 2). All 13 patients require continuing follow-up with a pediatric cardiologist.

Children Requiring a Procedure (n = 6)

A secundum atrial septal defect with a left to right shunt greater than 2:1 was found in 2 children. In both cases, typical auscultatory findings, consisting of a widely and fixedly split second heart sound, a systolic ejection murmur in the left upper sternal border and diastolic rumble at the right lower sternal border were present. One patient underwent surgical closure of the defect and the other awaits trans catheter device closure.

A small patent ductus arteriosus was found in a child with a nonspecific systolic murmur and otherwise normal cardiovascular examination. He underwent cardiac catheterization with successful coil occlusion of the ductus arteriosus. Bacterial endocarditis prophylaxis was discontinued 6 months later, after documenting the lack of a residual shunt by color Doppler.

A small fistulous connection between the left main coronary artery and the right pulmonary artery was detected in 1 patient. Physical examination was normal. The patient underwent cardiac catheterization, which confirmed the echocardiographic diagnosis. Conservative follow-up was recommended.

Evidence of pulmonary hypertension of moderate degree was found in 1 child. Physical examination was unremarkable. Cardiac catheterization was recommended, but the patient declined. A quantitative pulmonary perfusion scan was not performed.

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axis and apical 4 chamber views and color Doppler evidence of trivial to mild (1+) mitral regurgitation were present in 3 of 4 cases. A mid-systolic click was heard in 2 of the children, and a late systolic murmur in the apex was auscultated in the third. In the fourth child, there was no definite prolapse, but the mitral valve appeared myxomatous and showed mild (1+) regurgitation; the same patient also had a patent foramen ovale with left to right shunt and no abnormal auscultatory findings.

A bicuspid doming nonstenotic aortic valve was diagnosed in 2 participants. A systolic ejection click and a systolic ejection murmur in the aortic area were auscultated in 1 child, and the other had trivial aortic regurgitation by Doppler with a normal cardiac examination.

An embryonal/spongy appearance of the left ventricular myocardium suggestive of cardiomyopathy was present in 1 participant.

Antibiotic prophylaxis for bacterial endocarditis was started in 8 children at the time of diagnosis. Based on current recommendations, this will be a lifetime precaution in 7. Bacterial endocarditis prophylaxis was discontinued in the 1 child who underwent coil occlusion of a patent ductus arteriosus 6 months later, after confirming the absence of a residual shunt by color Doppler.

**DISCUSSION**

Estimates of the prevalence of cardiac disease in a population are dependent on the methods used to detect and diagnose the cardiac lesions. Children with prominent physical signs and symptoms, eg, cyanosis, congestive heart failure, or loud murmurs, are quickly recognized and aggressively evaluated. In contrast, cardiac disease in asymptomatic children with inconspicuous physical findings may be easily missed during clinical examination, despite the presence of significant abnormalities.

This was the case in the present study in which nearly 4% of a randomly selected population of junior high school students was found to have previously undetected cardiac disease, severe enough in about half of them to require cardiac catheterization and/or surgery at the time of diagnosis. Cardiac disease had not been previously recognized in these children because of a lack of symptoms and a failure of the primary care givers to notice abnormal findings on cardiac auscultation. It is not surprising that noncardiologists would fail to detect subtle auscultatory findings. However, some heart lesions remain undiagnosed after physical examination by well trained specialists, and, as the present report shows, only 7/13 (54%) patients were found to have an abnormal cardiac examination by a pediatric cardiologist.

These data are consistent with a recent report showing that history and physical examinations performed by trained pediatric cardiologists are accurate in only 81% of cases after an initial evaluation and that significant cardiac disease can be missed without the use of additional diagnostic methods. It has been shown that echocardiography greatly improves the accuracy of the cardiac evaluation in children with mild disease. For example, the incidence of small muscular ventricular septal defects was found to be 53.2/1000 in consecutive neonates studied by Doppler echocardiography in contrast to the substantially lower incidence of 20/10,000 live births reported in earlier studies. The results of the present report are also in agreement with an earlier report showing that the use of echocardiography improved the detection of cardiac abnormalities in young children by 5%.

There are a number of reasons to justify earlier diagnosis of cardiac abnormalities. First, repair of congenital lesions, such as atrial septal defect, reduces the later risk of arrhythmias, right ventricular dysfunction, and pulmonary hypertension. Second, not only severe, but also some mild cardiac lesions, have potential long-term hemodynamic consequences and risk of bacterial endocarditis. The reports from the First and Second Joint Study on Natural History Studies of Congenital Heart Defects confirm an incidence of bacterial endocarditis between 0.03% and 0.06% patient-years, the highest rates corresponding to aortic valve abnormalities and advanced age of diagnosis. Third, diagnosis of either severe or mild defects is important for purposes of genetic counseling. The risk of recurrence of congenital heart defects has been reported to range between 1% and 15% after specific chromosomal genetic syndromes are excluded.

**CONCLUSION**

In summary, the findings of the present study suggest that a significant percentage of the childhood population has cardiac disease undetected by clinical means alone. Some of these children require corrective intervention procedures, prophylaxis for bacterial endocarditis, and/or follow-up by a pediatric cardiologist. The challenge is to develop a strategy to detect these children, because careful physical examination performed by a pediatrician may not be adequate. Although Echo-Doppler examination is highly sensitive and specific, the cost for this procedure limits its feasibility as a screening method under most current health care systems. Screening by a pediatric cardiologist successfully diagnosed abnormalities in half of a group of patients who were previously thought to be normal. Therefore, in a circumstance in which there is a question about abnormality of physical findings, screening by a trained pediatric cardiologist seems the reasonable primary method of evaluation. Nevertheless, even with screening by a pediatric cardiologist nearly half of the abnormalities would not have been recognized. Confirmation of these results in larger groups of children and developing an improved method of screening, more cost-effective, and less demanding than undirected echocardiography, should be addressed in future studies.

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REFERENCES


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