Health Supervision for Children With Down Syndrome
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Health Supervision for Children With Down Syndrome

To the Editor.—

The AAP’s Committee on Genetics recently published guidelines concerning the health supervision for children with Down syndrome. Although they recommended echocardiographic studies during the neonatal period, an echocardiogram was not recommended during adolescence and early adulthood. We believe that this is a significant omission.

In 1993 we reported on the development of valve dysfunction in adolescents and young adults with Down syndrome without any previous history of congenital heart disease. Thirty-five patients were studied and 46% had mitral valve prolapse; 2 of these patients also had tricuspid valve regurgitation. Two patients had aortic regurgitation. Another finding was that 4 of the 23 patients older than 18 years of age had valve regurgitation, but none of the 12 patients younger than 18 had valve regurgitation.

As a result of these findings, we recommended routine screening of adolescent and young adults with Down syndrome, especially before dental or surgical procedures, for the development of valve dysfunction. In the 8 years that we have followed this recommendation, we continue to uncover the presence of valve dysfunction in these patients, many of them without any cardiac findings on physical examination.

Therefore, we believe that routine echocardiographic studies should be a part of the health supervision of adolescents and young adults with Down syndrome.

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REFERENCES


To the Editor.—

We read the interesting and very useful guidelines of the Committee on Genetics of the American Academy of Pediatrics recently published in Pediatrics.1

The authors correctly suggest performing cardiac evaluations by a pediatric cardiologist in newborns with Down syndrome. Echocardiogram is recommended to rule out congenital heart disease because of the high prevalence (50%) of cardiac malformations in these children. However, the suggested guidelines do not include other cardiac evaluations for children, adolescents, and adults with Down syndrome.1

Unfortunately, some adolescents or young adults with trisomy 21, also in the absence of the classic congenital cardiac defects, may present some cardiac abnormalities characterized by late clinical presentation. These cardiac abnormalities consist of mitral valve prolapse (MVP) and aortic valve insufficiency.2–4 The prevalence of these abnormalities is not completely defined, but it was suggested that MVP may occur in 44% to 57% and aortic regurgitation in 11% to 14% of adults with Down syndrome.2–6 These abnormalities, not evident in neonatal age and in infancy, can be caused by weakness or laxity of connective tissue in agreement with the general ligamentous laxity in individuals with Down syndrome.6

The clinical relevance of these abnormalities is variable: in the majority of patients the defects are mild without major clinical signs, but some patients may present significant emodynamic consequence and request surgical treatment.

Our personal experience consists in 6 subjects with Down syndrome (age 13–22 years), in whom cardiac evaluations performed in infancy did not reveal congenital heart defect, but showed, during adolescence, clinical and echocardiographic signs of aortic regurgitation (2 cases) and MVP with trivial insufficiency (4 cases). One of these patients, who had severe aortic regurgitation, underwent aortic valve replacement at 16 years of age.
Mild forms of MVP and aortic regurgitation deserve particular attention before any form of surgical intervention, including dental procedures. Antibiotic prophylaxis is always indicated in these patients.

Recent studies on children and adolescents with Down syndrome without congenital heart disease report a good cardiac function and an adequate cardiopulmonary response to exercise test.

However, the possible late presentation of MVP or aortic regurgitation suggests that the guidelines for health supervision for adolescents or young adults with Down syndrome should include a second cardiac evaluation if the first assessment performed in neonatal age or in infancy did not reveal cardiac defects.

**In Reply.**

As the letters’ authors point out, the prevalence of cardiac abnormalities is substantial in adolescents and adults with Down syndrome without a history of congenital heart defects. It is less clear what the role of routine echocardiography should be in such a population. The publication by Geggel et al found that 18 of 35 patients (57%) had MVP, though none had mitral regurgitation. Of most concern in this report was the finding of aortic regurgitation in 4 patients who would not be detected clinically because they did not have a cardiac murmur. Antibiotic prophylaxis against endocarditis is indicated for this group before surgery. Marino et al’s personal experience with 6 patients is also used to support a recommendation for routine echocardiographic evaluation. However, their patients are described as having both clinical and echocardiographic signs of valve disease and therefore should be identifiable on physical examination.

In summary, there are abundant data that document the increased prevalence of cardiac valve disease in adolescents and adults with Down syndrome. However, the data are less clear regarding the most reasonable approach to screening this population. It appears that the great majority of these abnormalities can be detected through careful cardiac auscultation. The policy statement on “Health Supervision for Children With Down Syndrome” is meant to serve as a supplement to the American Academy of Pediatrics’ “Recommendations for Preventive Pediatric Care,” which calls for annual evaluations, including history and physical examination. For most individuals with Down syndrome and valve abnormalities, this screening would appear to suffice. Individuals with Down syndrome should certainly be considered at higher risk than the general population for valve abnormalities and receive a prompt referral for any suspicious findings on history or physical examination. A referral for echocardiographic evaluation should also be considered for individuals with Down syndrome who are to undergo surgery, but the need for routine echocardiography is not well-supported from the available literature.

**REFERENCES**


**Screening for Retinopathy of Prematurity**

**To the Editor.**

Current guidelines for the examination of premature infants for retinopathy of prematurity (ROP) suggest that infants with a birth weight <1500 g or with a gestational age of 28 weeks or under, or those infants with an unstable clinical course felt to be at high risk by their attending pediatrician or neonatologist, be screened.1 It is our understanding that revisions to these guidelines are currently under consideration. We present the following case report involv-