

This Month In **The JOURNAL** of **PEDIATRICS**

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We repair their hearts—but ignore their development

— Reginald L. Washington, MD

The fields of Pediatric Cardiology and Pediatric Cardiac Surgery continue to successfully prolong the lives of children born with congenital heart disease (CHD). The medical home for these children usually centers on a primary care provider who has the responsibility for their overall health and well-being.

Much has been learned regarding the neurodevelopment of these patients. In fact, specific guidelines have been published by the American Heart Association (2012) targeting the identification and management of developmental disorders in children with CHD. The goal of these guidelines was to optimize the care of children within their medical home by identifying those who are at the highest risk of developmental abnormalities. So how are we doing?

In this volume of *The Journal*, Knutson et al show that an opportunity exists to improve the use of these guidelines in the primary care setting. The pediatric community is not aware of the American Heart Association's guidelines and, therefore, does not use them as a tool to screen or refer children who would potentially benefit from testing and/or intervention. This report reviews barriers that inhibit the use of these guidelines, as well as presents easy suggestions that would facilitate putting the guidelines in the hands of primary care providers. Communication between the cardiology and the primary care communities needs to improve so that the overall well-being of these children is optimized.

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What did the Task Force say?

— Denise M. Goodman, MD, MS

This volume of *The Journal* includes a Commentary on the recent recommendations of the US Preventive Services Task Force (USPSTF) (hereafter "the Task Force") on universal screening for autism spectrum disorders. The authors give a comprehensive argument as to why they disagree with the Task Force's recommendations, and why universal screening is warranted.

To put this into context, it is useful to understand the charge and process of the Task Force, and how recommendations are graded. The Task Force was created in 1984 as an independent, volunteer expert panel in prevention and evidence-based medicine. The specific focus is to make recommendations on screening, counseling, and preventive medications, focusing only on people who have no signs or symptoms of the disease that the screening or intervention targets. The Task Force reviews existing studies, but does not conduct its own studies, and panel members are publically listed on the USPSTF web site, serving 4-year terms. After a topic is selected, a draft research plan is posted for public comment, and comments are incorporated into the final research plan. The Evidence-Based Practice Center then gathers, reviews, and analyzes evidence using the research plan as a road map, with the evidence summarized and graded. The draft recommendations and evidence review are also posted for public comment before final recommendations are made (<http://www.uspreventiveservicestaskforce.org/>, accessed May 31, 2016).

The statement on screening for autism spectrum disorder in young children reads, "The USPSTF concludes that the current evidence is insufficient to assess the balance of benefits and harms of screening for autism spectrum disorder (ASD) in young children [aged 18-30 months] for whom no concerns of ASD have been raised by their parents or a clinician", and was given an evidence grade of "I," which means that "The

USPSTF concludes that the current evidence is insufficient to assess the balance of benefits and harms of the service. Evidence is lacking, of poor quality, or conflicting, and the balance of benefits and harms cannot be determined.” The reader is prompted to read the clinical considerations section of the Recommendation Statement, with the conclusion, “If the service is offered, patients should understand the uncertainty about the balance of benefits and harms.”

Echoing the Commentary, the clinical considerations sections acknowledges the benefits of early intervention; reiterates the burden of ASD on patients, families, and the healthcare system; recognizes that racial/ethnic and socioeconomic disparities exist with respect to diagnosis; and lists potential harms as principally related to misdiagnosis and anxiety pending confirmatory testing.

So, where is the controversy? Unfortunately, to many readers, “we don’t know” is misinterpreted as “don’t perform screening.” Although public health has become regrettably politicized, the important message is that ASD is a significant public health concern, screening can easily be accomplished in a primary care setting, and we need sufficient behavioral health resources both to address concerns raised by preliminary screening and to perform early intervention in those for whom a diagnosis of ASD is confirmed.

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Why it is important to assess hypermobility in very young children

— Philip J. Hashkes, MD, MSc

Hypermobility is associated with certain functional benefits (eg, sports and music), but with many potential deleterious consequences, some of which have been reported in previous volumes of *The Journal*. These include joint and muscle pain, “growing-like” pains, gastrointestinal symptoms, and even fibromyalgia, which can substantially impact the quality of life of affected children. A key potential consequence in younger children is developmental delay, particularly in gross motor milestones. Therefore, early detection of hypermobility is of importance. Prior to this study, objective diagnostic criteria were defined only in school aged and older children.

Romeo et al adapted the most commonly used criteria, the Beighton score, for use in toddlers and preschool children. The major change they made was to substitute back flexion with ankle dorsiflexion, because it is difficult to request a very young children to perform a reliable back flexion. A surprising finding of the study was that the cut-off score the authors found to define hypermobility (>4) in their preschool cohort was similar to the accepted score in older children. Per clinical experience, it was expected that younger children would be much more flexible than older children. The results were confirmed in a group of children with syndromes associated with hypermobility. A limitation of this study is the need to prospectively follow these young children to observe changes in their flexibility as they grow. This study is a first step in defining hypermobility in very young children. Early screening for hypermobility may lead to greater vigilance in following developmental milestones and to early referral for physical therapy intervention, if needed. It also would be of interest to study whether early intervention may prevent some of the later deleterious consequences of hypermobility.

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Genes and patent ductus arteriosus

— Stephen R. Daniels, MD, PhD

The genetics of cardiovascular disease has been a confusing area, in part because our classification of cardiac morphology has not mapped well to genetic influences on cardiac development, but also because cardiac development is a very complex process involving many genes and environmental influences.

In this volume of *The Journal*, Patel et al evaluate single nucleotide polymorphisms in candidate genes potentially associated with patent ductus arteriosus (PDA) in term infants. The candidate gene approach can be troublesome because it is often difficult to know which genes to pick. When results are negative, it is not always clear whether the wrong genes were chosen, the power was inadequate, or if there were other issues. When the results are positive, it is not always clear whether there is a causal relationship or whether significance (if not corrected) resulted from multiple statistical tests.

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