Letter to the Editor Pediatrics AAP 2011

Health Supervision for Children with Down Syndrome 2011- Revisited

Commendations to the AAP *Committee on Genetics* for its recent work to update the guidelines for "Health Supervision for Children with Down Syndrome" (Bull 2011). For pediatricians serving children with Down syndrome (DS) these guidelines represent a much anticipated advancement over the previous guidelines published in 2001 (Cunniff 2001). The revised document succeeds in articulating the complex health manifestations associated with DS and serves well to educate and guide pediatricians and other primary care providers.

Organizing health recommendations according to age-epoch allows for the presentation of disparate information in a straightforward manner which is useful for monitoring children with complex conditions. The guidelines emphasize the importance of monitoring, screening, and diagnosing those medical conditions known to result in significant morbidity or mortality in this population. The guidelines do not attempt to provide management or treatment decision algorithms, which are instead deferred to the judgment of providers. Suggestions for anticipatory guidance which address development, behavior, social-emotional well-being and child-family adjustment are provided to anticipate and plan for future needs. Access to current, balanced sources of information and local community-based services for children and families is emphasized. The committee has also expanded the recommendations on prenatal care for expectant couples.

New recommendation highlights from the current guidelines include: obtaining a radiographic swallow study on neonates and infants with severe feeding problems or respiratory complications; a car safety seat evaluation for neonates with apnea, bradycardia or oxygen desaturation; monitoring hemoglobin annually and serum ferritin as indicated for iron deficiency; obtaining an overnight polysomnogram on all children by 4 years of age due to the high incidence of sleep-related breathing disorders; performing a screen for celiac disease in symptomatic individuals exposed to a gluten containing diet; and obtaining an echocardiogram in adolescents and adults with symptoms of acquired aortic or mitral valve disease.

Some familiar recommendations have been retired: use of the currently available, but outdated DS growth curves, has been discontinued in favor of using standard NCHS or WHO growth charts in conjunction with standard measures of body mass index (BMI) until updated DS growth charts become available. It isn't clear that outright abandonment of the older DS growth curves, in favor of the standard instruments, provides any obvious advantage for monitoring growth in young children. Use of both the standard and currently available DS growth charts may provide some reassurance for healthy children with DS who grow at a steady rate but remain below the 3rd percentiles on the standard curves. Routine cervical spine radiographs at 3 years have been discontinued in favor of aggressive monitoring for symptoms of myelopathy related to cervical spine instability. Spine radiographs and further evaluation are recommended only for symptomatic individuals. Introduction into the guidelines of the term "myopathy" rather than consistent use of "myelopathy" appears to be a typographical error which may cause some confusion among providers assessing neurological function.

It is not always clear how the *Committee* chose to distinguish recommendations that warrant *evaluation or assessment* (requiring examination, laboratory studies or specialty referral) from those listed under *anticipatory guidance* (requiring discussion, counseling or review). Several of the recommendations listed under anticipatory guidance call for the provision of service (i.e., provide influenza and/or PPS23

vaccines annually) or the need for regular assessment for autism and behavioral problems. These particular recommendations as guidance are indistinguishable from those requiring assessment or evaluation. Behavioral concerns are, after all, common in children with DS (Dykens, Shah et al. 2002) (Capone, Goyal et al. 2006), and we agree that regular assessment and prompt referral is indicated when problems are present. If recommendations for assessment are made based on the strength of available studies to support such recommendations, this presumption has been left unstated. Indeed, the methodology for literature review, selection and the grading of evidence has not been articulated.

One of the many challenges inherent to developing health supervision guidelines for children with complex medical needs is how to make a diverse and expansive set of recommendations accessible to pediatricians practicing across a variety of ~~s~~settings. For clinicians unfamiliar with the needs of children with DS the document can seem daunting by its expectation of limitless time, patience, wise counsel, and persistent requirement for sustained vigilance. In the primary care setting the measured benefits of anticipatory guidance have been questioned as the number and complexity of recommendations for preventive care has expanded (Needlman 2006). In contrast children with DS represent a high-risk population so the impact of parent education and anticipatory guidance as "preventive medicine" may have greater significance, although this remains untested.

Among the community of physicians serving individuals with DS there is a compelling need for more robust guidelines to address the screening and diagnosis of specific behavioral and mental health conditions, and those with multiple medical co-morbidities which affect the physical and mental health of individuals with DS of all ages. Finally, clinical research studies to inform the design of future evidence-based practice guidelines are sorely needed (McCabe, Hickey et al. 2011).

As evidence about the health conditions of children with DS continues to expand, the development of future guidelines is likely to prove increasingly challenging. The process will always require the proper balance of evidence-based support and consensus-based expert opinion because both contribute to the implementation of best practice. When expert opinion serves as the basis for a recommendation it will be useful to understand the supporting rationale. In future iterations the methods used to guide the literature review and grading of evidence needs to be formalized. If scientific evidence is lacking or weak, then it needs to be articulated along with suggestions about how to generate such evidence. Adopting this style of dialogue will promote a higher standard of care for individuals with DS and help to focus clinical research efforts on their behalf.

For the Down Syndrome Medical Interest Group

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