Clinical report updates diagnostic criteria for Marfan syndrome

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The revised AAP clinical report Health Supervision for Children with Marfan Syndrome incorporates the revised Ghent diagnostic criteria and updates several key clinical areas.

This long-awaited update from the AAP Committee on Genetics is designed to assist pediatricians in recognizing the features of the syndrome and caring for patients. It replaces the 1996 guidance and is published in the October issue of Pediatrics (2013;132:e1059-1072).

Use of a systemic score

Marfan syndrome is a heritable, multisystem disorder of connective tissue with extensive clinical variability. It is a relatively common condition, with approximately one in 5,000 people affected. About 25% of cases are sporadic, but some familial cases go unrecognized. Often, it is the pediatrician who is central to the diagnosis and ultimately the management of this condition. The pediatrician often recognizes features concerning for Marfan and refers for further evaluation, which may be cardiac or genetic.

The original Ghent diagnostic criteria used some features that had not been validated sufficiently, were not applicable to children, or necessitated expensive and specialized investigations. The newly revised diagnostic criteria from an international panel of experts are meant to simplify the diagnosis of Marfan syndrome with increased specificity but still rely heavily on family history or proven genetic mutation, dislocated lenses (ectopia lentis) or a dilated aorta. The various physical and radiologic criteria have been simplified somewhat in a systemic scoring system — and yes, there is an app for that (see resource).

Coordination of care

Once diagnosis is established, management becomes multidisciplinary. Some patients are able to take advantage of specialized clinics, but many are not. Coordination of care becomes the family’s responsibility, which can be a burden that many pediatricians take on.

Patients need cardiology and ophthalmology follow-up. They need to be monitored for musculoskeletal issues such as scoliosis, pectus deformity or joint laxity, which may require different specialists. Various medications may be prescribed, and it becomes necessary to review these frequently. Often, families have questions about activities, including physical education classes, as well as growth and possible hormonal treatment to stunt excessive growth. Health care professionals also must be cognizant of the patient’s emotional issues, including body image concerns, activities, sexuality, reproductivity and mortality.

Improvements in treatment

The latest information on the treatment of Marfan syndrome also is described in the clinical report. Much progress has been made in aortic replacement, especially with valve-sparing techniques that don’t require lifelong anticoagulation. Similarly, lens stabilization surgeries or implantable lenses offer more options to those affected with Marfan syndrome.

One of the more exciting prospects is the use of angiotensin-receptor blockers that not only may prevent aortic dilatation but reverse what already has occurred; however, studies still are ongoing.

Dr. Tinkle is a lead author of the clinical report.
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