



National Marfan Foundation • 1981-2011

National Marfan Foundation Launches Mobile Site to Put Marfan Syndrome Diagnosis Directly into the Hands of Physicians

Differential Diagnosis and Management of Related Conditions also Featured on www.MarfanDX.org

PORT WASHINGTON, NY, April 15, 2011 -- A new mobile site – www.MarfanDX.org – was launched today by the National Marfan Foundation (NMF), to facilitate diagnosis of Marfan syndrome and related disorders. The site content is based on the new diagnostic criteria for Marfan syndrome that were published in the *Journal of Medical Genetics (J Med Genet 2010;47:476-485)*. The site is compatible with Droid and iPhone smartphones.

“The new mobile site is an innovative use of new technology for doctors who are concerned about the possibility of underlying Marfan syndrome or a related connective tissue disorder,” said Mary Roman, MD, Professor of Medicine, Weill Cornell Medical College, and a member of the NMF Board of Directors. “Marfan syndrome is a condition that has a varied expression and requires a multi-system clinical examination for diagnosis. The revised criteria simplified the complex process for diagnosis and provided insights into the diagnosis for conditions with overlapping features. The NMF’s new mobile site facilitates instantaneous access to diagnostic criteria for Marfan syndrome and provides diagnosis and management information for related disorders. This is a real benefit for busy doctors.”

Marfan syndrome and related disorders affect about 200,000 people in the U.S., but experts say that half of those affected are not diagnosed. Without a diagnosis and treatment, they are at risk of a sudden early death from a tear or rupture of their aorta. Related disorders include Loeys Dietz syndrome, vascular Ehlers Danlos, mitral valve prolapse syndrome, familial aortic aneurysm and more, are outlined.

Features of the Mobile Site

According to a study conducted last year by Manhattan Research, a full 72 percent of U.S. physicians now use smartphones, and the number is expected to jump to more than 80 percent by 2012.

“With so many physicians utilizing smartphone technology,” says Dr. Roman, “it makes sense to provide a resource that is compatible with the latest trends.”

The mobile site features a summary of the new diagnostic criteria, including seven simple formulae for diagnosing Marfan syndrome. Expandable text provides a detailed explanation of each formula.

The site also features:

- Interactive Systemic Score Calculator used to consider the lesser characteristics of Marfan syndrome throughout the body that can be key in making the diagnosis. This too has expandable text and graphics, as well as ability to email results for patient file.
- Interactive Z-score calculator, used to determine the size of the aorta compared to body surface area. This can also be emailed for the patient file.
- Key points about the role of genetic testing and family history.
- Important information on differential diagnosis and related disorders.
- Helpful links and resources.

The mobile site is also viewable on a desktop computer; it is compatible with Safari and Firefox browsers.

This mobile web site was supported by the CDC Cooperative Agreement Number 1H75DD000703-01. Its contents are solely the responsibility of the authors and do not necessarily represent the official views of the Centers for Disease Control and Prevention.

Revised Diagnostic Criteria for Marfan Syndrome

The revised diagnostic criteria for Marfan syndrome were developed by an international team of medical experts led by Bart Loeys, MD, Center for Medical Genetics, Ghent University Hospital, Ghent, Belgium. The purpose was to streamline the diagnostic process in order to provide patients with a more accurate diagnosis and better medical management.

"While diagnostic criteria should emphasize simplicity of use and the desire for early diagnosis, the highest priority in developing these guidelines was accuracy," said Dr. Loeys.

Because connective tissue makes up the entire body, the disorder manifests itself in many body systems, including the skeletal system, eyes, lungs, blood vessels and heart. Many people with Marfan syndrome experience an expansion of the aorta. It is essential for affected people to be diagnosed and managed properly. Without proper monitoring and medications to reduce the stress on the aorta, affected people are at high risk for aortic dissection or rupture, which could result in sudden death. With an accurate diagnosis and proper medical treatment, they can live a normal lifespan.

The revised nosology provides a method for evaluating a patient by deriving a systemic score, with various features of Marfan syndrome assigned a numeric value; the diagnosis depends on the total systemic score. This is a change from the previous nosology which relied on evaluation of features as "major" or "minor."

The scoring system reflects three significant changes in the way Marfan syndrome is diagnosed:

- The two cardinal features of Marfan syndrome - aortic root dilatation/dissection and ectopia lentis (dislocated lens of the eye) – are weighted more heavily than other characteristics.

- There is a more precise role for molecular testing.
- Less specific manifestations of Marfan syndrome are either removed or given much less weight in the evaluation process.

The diagnostic criteria have been defined for those with a family history of the condition and for those who may be a sporadic case; that is, they are the first in their family to be affected. Specific guidelines are also given for children (less than 20 years of age), with different scenarios proposed for those with family history and those without family history. For those who do not meet the diagnostic threshold for Marfan syndrome or a related condition, the nosology employs the diagnosis of "non-specific connective tissue disorder," which fosters ongoing monitoring of the aortic size and function, until such a time when a specific diagnosis can be made.

The nosology also offers additional diagnostic considerations and recommends more testing if a patient has sufficient findings of Marfan syndrome but, additionally, shows other unexpected features. The differential diagnosis and management for alternative diagnoses, such as Loeys Dietz syndrome, vascular Ehlers Danlos, mitral valve prolapse syndrome, familial aortic aneurysm and more, are outlined.

The National Marfan Foundation

The National Marfan Foundation is a non-profit voluntary health organization dedicated to saving lives and improving the quality of life of individuals and families affected by the Marfan syndrome and related disorders by:

- Educating affected individuals, family members and the health care community about Marfan syndrome.
- Advocating for and funding clinical and molecular research into the early detection and treatment of Marfan syndrome.
- Providing a network of local and special-interest support groups to help affected people and their families share experiences.

For more information on the Marfan syndrome, call 800-8-MARFAN or log on to www.marfan.org.

* * *