

Study describes symptoms and severity of fibromuscular dysplasia

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A patient registry is proving invaluable in cataloging the clinical features, symptoms, severity, and outcomes of fibromuscular dysplasia (FMD), a non-inflammatory vascular disease that can cause narrowing of arteries in the carotid (neck) and renal (kidney) arteries, which can result in headache, strokes, and aneurysms. Registry data revealed that 91 percent of patients are women diagnosed in the prime of their lives. As a result, physicians are developing best practices that can lead to early diagnosis and treatment with the goal of improved outcomes and fewer adverse vascular events. The registry results are published in the June 26, 2012 print edition of *Circulation*.

"This is the largest study published to date; most previous studies included single case reports or small case series," says Jeffrey W. Olin, DO, the study's lead investigator, and Professor of Medicine and Director of Vascular Medicine and the Vascular [Diagnostic Laboratory](#) at Mount Sinai School of Medicine. "Seventy percent of the people in the registry have [high blood pressure](#), 10 percent have had strokes, and 20 percent have had an [aneurysm](#)." Additionally, there was an average delay in diagnosis of approximately four years from the onset of symptoms.

The study reviews the first 447 patients enrolled in the FMD registry at nine centers in the United States, including Mount Sinai. The registry has since grown to nearly 600 patients. Until now, the disease has been considered rare, but Dr. Olin thinks that it is probably much more common, and some experts believe it may affect up to 4-5 percent of the

female population.

Signs of FMD that physicians and patients should be attentive to include:

- high [blood pressure](#) in patients under age 35, or uncontrolled high blood pressure at any age;
- unrelenting headaches;
- pulsatile tinnitus, where a sound in the ear sounds like a swooshing and occurs with the heartbeat;
- stroke under age 60;
- a bruit (abnormal sound when listening with a stethoscope) in the abdomen;
- a noise in the neck when the doctor listens to it, called carotid bruit, suggesting there is turbulence to blood flow in the artery;
- a dissection or aneurysm of an artery.

A big challenge to diagnosing FMD continues to be that many doctors do not listen to the neck with a [stethoscope](#), says Dr. Olin. Diagnosis is confirmed through imaging, such as ultrasound, CT angiography, MR angiography, or catheter-based angiography.

The cause of FMD is unknown. Treatment varies. For example, when FMD results in high blood pressure, percutaneous balloon angioplasty may be performed. For a tear in an artery (dissection), physicians may recommend stenting the affected artery. In some cases, anti-platelet drugs such as aspirin may be used. Still, investigators say more data are needed to understand the cause of the disease and to treat it optimally.

"If the field is to move forward, we urgently need more funding for genetic research into patients affected and other family members," says Dr. Olin.

Provided by The Mount Sinai Hospital

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