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## The 'Rare' Disease That Isn't

*Often Undiagnosed, FMD May Afflict Up to 5% of Americans*

By [THOMAS M. BURTON](#)

It took an autopsy to determine why 10-year-old Haley McWhorter didn't wake up one morning last May.

While asleep, Haley went into cardiac arrest, stopped breathing and never started again, concluded the medical examiner in Ft. Myers, Fla. The examiner found that a thickened artery wall had blocked blood flow to Haley's heart. The odd growth of the artery wall suggested the presence of a disease called FMD, or fibromuscular dysplasia, the examiner concluded.

FMD, a condition in which artery walls expand into and obstruct the arterial channel, is largely unknown to the public and even to the majority of doctors. When discussed in medical schools -- if discussed at all -- FMD is typically described as an obscure and rare disease.

Yet a tantalizing body of evidence has begun to emerge that suggests FMD isn't rare at all: It simply isn't looked for, so it is seldom diagnosed.

Some FMD patients aren't diagnosed even after arriving in doctors' offices with such severe events as strokes; burst aneurysms, or ballooned sections of arteries; and artery dissections, in which the inner artery lining peels away and hampers blood flow. Some FMD patients say they get dismissed by doctors who, rather than admit they don't understand what is wrong, tell patients that their problems are psychosomatic.

A few thousand cases of FMD have been confirmed in the U.S., mostly during the last decade. The National Stroke Association in 2005 listed FMD as a cause of strokes. This year, a group of vascular specialists across the U.S. started a computerized registry of patients to analyze FMD's scope, causes and treatments.

"I believe that a large number of Americans have FMD," says Jeffrey W. Olin, director of vascular medicine and a professor at the Mt. Sinai School of Medicine in New York City. "It's reasonable to say that many thousands could be saved from complications like heart attack, stroke, ruptured aneurysm and even death, by screening patients."

There have been clues to FMD's scope for years. In one series of random consecutive autopsies at the Mayo Clinic in Rochester, Minn., in the 1970s, nine, or more than 1%, of 819 patients had FMD. In a separate, 1989 study of 1,862 patients who volunteered to donate kidneys, 71, or 3.8%, were found through scans of their renal arteries to have FMD. This year in the *European Journal of Radiology*, a study of 101 potential kidney donors found an almost identical rate -- four patients, or 3.9%.

"Three to five percent would be a very reasonable estimate in the general population for FMD," says Thom W. Rooke, vascular medicine professor at the Mayo Clinic. That works out to there being possibly 10 million Americans who have FMD, he says. By comparison, an estimated 3 million Americans have epilepsy, 2.5 million have breast cancer, more than 2 million have schizophrenia and 725,000 have melanoma.

Dr. Rooke says many FMD patients might never have symptoms, and only a few are likely to experience severe problems. But, he says, many "vascular catastrophes," such as heart-rhythm deaths and strokes in young people, may

stem from FMD.

About 85% of the known FMD cases have affected women under 50 and girls, but boys and men also get it. It is not clear how the disease originates, though one French study reports that more than 10% of patients have close relatives with FMD. Dr. Olin says it is more like 5% in his patients. Last year, the National Institutes of Health identified several genes possibly linked to FMD.

FMD was first described in a medical journal in 1938, but until roughly the past decade, publications described only isolated, individual cases. Vascular-medicine doctors, who specialize in diseases of the arteries and veins, generally say they received no information about it in medical schools. The field of vascular medicine itself is relatively small, with only a few hundred doctors, plus over 2,000 surgeons, practicing in the U.S. In contrast, there are roughly 25,000 cardiologists.

Research on FMD is scant, partly because the pharmaceutical industry hasn't envisioned potentially large profits. Current treatments include blood-pressure and anti-clotting drugs, and angioplasty to open arteries.

Dr. Olin, 58 years old, began gravitating toward vascular medicine during a residency at the Cleveland Clinic in the early 1980s. Because the clinic is mostly a referral hospital, meaning doctors send patients there when they can't figure out what is wrong, he was exposed to obscure vascular diseases.

These days, he is known among colleagues as much for his FMD research as for his repeated wins at "Vascular Jeopardy," a trivia game played every year at a meeting of vascular specialists.

While still at the Cleveland Clinic in the early 1990s, Dr. Olin encountered a 29-year-old patient who made a lasting impression. Lori Gardner, vibrant and otherwise in good health, had had a series of mini-strokes, constant fatigue and headaches, and a sky-high blood pressure of 250 over 150. Her doctors, she recalls now, had told her she had "stress."

"I'd been through so many doctors who didn't have the time for me," she says. Dr. Olin performed scans of the carotid arteries in her neck and renal arteries, those that supply her kidneys, and found FMD. When FMD is present, the artery lining pushes into the artery channel, while adjacent artery sections often bulge out in an alternating pattern. On an imaging scan, an affected artery often looks eerily like a string of beads.

Dr. Olin told Ms. Gardner she had FMD. Scans of the renal arteries and carotid arteries in the neck wouldn't normally be done unless doctors were investigating other possible diseases, yet they are the most definitive way to diagnose FMD. High blood pressure in patients under the age of 35 is another red flag, say doctors.

Many vascular-medicine doctors say they weren't taught about FMD in medical school, but some schools say they address it. At the Yale School of Medicine, Deputy Dean for Education Richard Belitsky says FMD is included in a second-year class on the kidneys and during a third-year primary-care rotation.

By the late-1990s, Dr. Olin was publishing his findings about patients with FMD, including treatments that tended to work. As he published, more FMD patients kept arriving at his doorstep.

Others were referred by Pam Mace, an FMD patient herself. Now president of the FMD Society of America, Ms. Mace is an emergency-room nurse from the Cleveland area who lives today in Grosse Ile, Mich. Slim and athletic, Ms. Mace was running marathons until 2000. In July 2000, she awoke with a headache. She later looked in a mirror and noticed dramatically unequal pupils, which she knew could indicate a stroke. One side of her face was numb.

At the emergency room, Ms. Mace says, her blood pressure read a disturbingly high 210 over 130. But it took three visits to two hospitals to conclusively diagnose that she had dissections, or major tears, in the lining of arteries supplying her brain. Even then, FMD wasn't diagnosed and treated until the following year.

One of the patients Ms. Mace later referred to Dr. Olin was Bruce Tagg, 54. An insurance safety consultant from Allentown, Pa., Mr. Tagg was suffering severe pain in his left flank two years ago. He was vomiting, had a fever of 102 degrees for six weeks and showed high blood pressure. Doctors gave him antibiotics.

But he didn't have an infection. Even after an image showed the characteristic beading in one renal artery, doctors "totally missed a dissection" in the other, says Dr. Olin. "The kidney was infarcting, meaning it was dying," because the renal artery was blocked off. Now, after angioplasty, a process that pushes the walls of the artery back out using a tiny

balloon, Mr. Tagg is in better health, despite the death of extensive kidney tissue.

Even when FMD is correctly diagnosed, doctors can make mistakes with treatment. In October 2007, doctors found a blockage in Rochelle DesRochers's renal artery. She was 45 years old, and told she had FMD. She says she brought literature to her doctor showing that the use of stents for such a blockage was generally a bad idea with FMD. "We do this all the time," she recalls her doctor saying. He placed a metal stent in her renal artery, she says.

"Forty-five minutes into recovery, I started feeling pain," she says. Doctors found the artery had dissected, or torn, and was further blocking blood flow to the kidneys. They put in another stent to try to reopen the artery. Within days, despite being in a lot of pain, she was sent home by doctors. It turned out that the now-dissected artery had thrown a blood clot into her kidney. She now is being treated by Dr. Olin, who says a stent was the wrong procedure and that it probably caused the clot and death of extensive kidney tissue.

Not all patients get diagnosed during their lifetimes. Haley McWhorter, the 10-year-old whose autopsy suggested FMD, had dizziness and headaches in her final weeks. Her mother, Laura Lemus, says she took Haley to a pediatrician, who said there was no reason to suspect a severe illness. "He said it was nothing, and that I didn't have anything to worry about," says Ms. Lemus.

Dr. Olin says that while Haley's symptoms could have been signs of the disease, they also could have been interpreted as any number of other things. Ms. Lemus says Haley's siblings have been tested but no FMD was found.

In another case, Adam Nickel, 27, had suffered no symptoms before he collapsed March 1, 2008, after finishing a marathon in Little Rock, Ark., his mother says. Like Haley, he died of sudden cardiac arrest from FMD, says Stephen Erickson, the medical examiner who did the autopsy.

Dr. Olin has prepared a top-10 list of doctors' misconceptions and missteps. They include, "Telling patients that the symptoms are all in their head. Telling patients that FMD is a rare disease. Telling patients with severe headaches with FMD that there is nothing that you can do for them."

The signs of FMD are frequently the same: young patients with high blood pressure, or who have had a stroke or temporary symptoms of a stroke; patients whose blood makes a swishing sound indicating turbulent flow; or patients with brain aneurysms.

Last year, doctors started an FMD registry to compile basic information: How many patients have immediate family with the disease? Why do only some patients suffer strokes or artery dissections? Will a symptom-free patient necessarily stay that way?

The registry began enrolling patients in January. One, Michigan resident Shawn Haven, was first diagnosed at the Mayo Clinic by vascular specialist Robert McBane. The lesson from Ms. Haven's case: Some doctors don't think to use a stethoscope.

Ms. Haven, now 45, went to a doctor in Michigan in October 2007 with severe abdominal pain and high blood pressure. She was very thin and unable to gain weight.

By early 2008, she says, she had "horrific pain" in the stomach and went to the emergency room. Two doctors, she says, concluded her problems were in her head. She recalls one of them telling her, "Just look at you, you're trying to make yourself sick."

Ms. Haven and her husband flew to the Mayo Clinic, where a doctor put a stethoscope to her abdomen. He heard a whooshing sound that indicates artery blockage and turbulent blood flow. Soon after, Dr. McBane found that FMD was blocking arteries to Ms. Haven's upper intestine, spleen and liver. The upper-intestine blockage led to the pain.

Though the arteries have since been cleared, doctors say Ms. Haven continues to deal with symptoms from artery blockage. She says she has to stick to a bland diet and is in near-constant pain: "I am going to be living with this for the rest of my life."

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