

State of limbo



A definitive MS diagnosis can take time—and patience.

by Brandie Jefferson

For 12 years, Kathy Volk, PhD, has not just been in limbo, but “way in limbo,” she says. That’s because for more than a decade, she has been unwell. For more than a decade, she and her doctors have struggled, unsuccessfully, to understand how her symptoms—numb feet, a squeezing feeling in her core and extreme fatigue—came together in a single diagnosis.

At one point, after MRIs, a lumbar puncture, bladder surgery, medications and a neurologist who said he just didn’t know how to help, she asked him to refer her to another doctor. He said he would, but added, “I just don’t know who to refer you to.”

Eventually, the Maryland resident, wondering if she had multiple sclerosis, took it upon herself to see an MS specialist. She was under his care for a decade while also seeing other doctors for different conditions. Ten years later, one of her doctors diagnosed her with common variable immune deficiency. Her MS specialist suggested that the treatment for that condition might solve her mysterious health problems. It turned out that he thought she might not have MS, after all.

Searching for answers

In September 2017, around her 51st birthday, Kathryn Knapp came down with a bad cold. A week later, she had trouble feeling her feet. Knapp, who lives in Pennsylvania, had a family history of health problems, so she was health-conscious and active. She thought the problems might be related to a running injury.

An orthopedic specialist prescribed steroids. On a Sunday, Knapp felt better. The following

Thursday, she lost feeling below her waist. Knapp spent the next three weeks in the hospital. She couldn't walk. Doctors suspected transverse myelitis, inflammation of the spinal cord. After the results of a lumbar puncture came in, she finally had a diagnosis.

"I was told I have MS and I should treat it aggressively," she says. Knapp saw a second neurologist. "I was told I don't have MS." She saw a third neurologist. "He said, 'We're not sure.'"

A waiting period

Although there is no solid data regarding how many patients are in this state of diagnostic limbo, neurologists say it's not unusual. In fact, a period of waiting is often integral to making an accurate diagnosis of MS. This waiting can be scary and frustrating. For some patients, however, a diagnosis seems like it will never come, and it can elicit a more fundamental question: Why is it taking so long to figure out what's wrong?

An MS diagnosis is far from straight-forward, in large part because there is no biomarker for the disease. No single test can rule out or verify its presence. In principle, the diagnostic process is simple, laid out by the McDonald Criteria: a syndrome typical for MS, two or more lesions in the brain, spinal cord and/or optic nerve that occurred at two different points in time. Critically, also, any other causes that might be responsible for symptoms or lesions must be ruled out.



Andrew Solomon, MD, has studied common diseases and conditions that can be mistaken for MS. Photo courtesy of Dr. Solomon

In practice, however, a diagnosis can be much more complicated. It often takes time and persistence to make an accurate diagnosis of MS.

“Overall, MS is not always hard to diagnose, and we have diagnostic criteria that work well,” said Andrew Solomon, MD, associate professor of neurological sciences and division chief, Multiple Sclerosis at the Larner College of Medicine at The University of Vermont. “Yet people still get misdiagnosed because in the majority of patients, the diagnostic criteria were not being applied correctly. There are a lot of clinical judgments involved in making a correct diagnosis that can be prone to error. Right now, without a biomarker, the neurologist is the screening tool.”

Pinpointing MS

In 2007, Volk had just turned 54. She was hiking with her husband when all of a sudden, “I just wet myself. I didn’t feel it. It just happened,” she says. It kept happening. A gynecologist recommended bladder suspension surgery, a common procedure to cure incontinence.

After the surgery, both of Volk’s feet went numb. Her primary physician suspected Guillain-Barré syndrome, a disorder in which the immune system attacks the peripheral nervous system. Volk’s doctor told her to see a neurologist if it didn’t clear up soon.

She didn’t go right away.

“I didn’t want to see a neurologist,” she says. It wasn’t that Volk was afraid of a diagnosis. At the time, she didn’t even know what a neurologist might be looking for. “I wasn’t scared. I just thought, ‘Oh come on, this is crazy.’”

When she did see a neurologist, he suspected Volk had a virus. When the tests came back negative, he told her to follow up in a few months, but she returned sooner. Volk had woken up one night with a horrendous squeezing in her ribcage. The neurologist ordered a spinal MRI.

“I had no idea what he was looking for,” Volk says. “I thought maybe I had a pinched nerve.” She was confused and starting to worry. “None of this was going away. It was scary.” Her job involved driving around the state of Maryland, but she didn’t feel that she could keep it up.

“I couldn’t do it. It was way too much. My feet were a mess. My legs were a mess. I was scared.”

In the fall of 2007, she took medical leave from her job.

Volk had worked in education as a teacher and administrator for three decades and was about to apply for a professor position at a local private college. “I didn’t know what was going to happen,” she says.

There were more tests, no answers. A spinal tap that was, in Volk’s words, “very difficult.” No answers. Another MRI, this one of her brain. There were lesions in her brain, but still no

answers. What made it worse, Volk says, is that her neurologist never said what he was looking for, leaving her with a fear of the unknown. It was her primary care physician who ultimately suggested that all of the scans, the lumbar puncture and physical exams were looking for MS.

But Volk's symptoms never fully met the diagnostic criteria. "My doctors were very confused," she says, "and I was getting more scared."

A second and third opinion

Knapp's first neurologist treated her with Ocrevus, at the time a newly approved therapy for MS. "I was so happy to hear that we were going to treat it aggressively. That's what a patient wants to hear," Knapp says.

At the same time, Knapp said she didn't feel that her doctor was communicating with her as well as he should be. He didn't run any additional MRIs or tests but instead relied on the information from her stay in the hospital.

"I thought, 'This is not right.' And you have to go with your gut feeling," she says. She sought a second opinion.

Her second neurologist, after reviewing her MRIs, wasn't sure that Ocrevus was the right thing to do.

"He said, 'I'm not sure you have MS from your MRIs. I never would have gone with Ocrevus without definitely confirming the diagnosis,'" Knapp recalls. Part of that confirmation entailed ordering new MRIs, but it also meant sitting down and listening to Knapp give a detailed history of her symptoms, past and present.

There are many common diseases and conditions that can be mistaken for MS. In a small 2016 study, a team of researchers led by Dr. Solomon identified 110 patients who had been misdiagnosed with MS. The most common correct diagnosis was migraine, but patients also had conditions such as fibromyalgia, psychiatric disease and small vessel disease.

"There are many other rare syndromes that are tricky to diagnose and can mimic MS," Dr. Solomon says. "Only with time is it often possible to tell that it is something else."



Bardia Nourbakhsh, MD, notes that it's important to get treatment while gathering clues for a diagnosis. Photo courtesy of Dr. Nourbakhsh

So Knapp and her doctors began the integral step of ruling out other possible causes of her symptoms. Her initial MRI revealed an issue with a disc, so she had neck surgery. After multiple MRIs, a lumbar puncture, steroids, starting and stopping Ocrevus, and then surgery, Knapp was ready for a third opinion. She traveled from Pennsylvania to Baltimore to see Bardia Nourbakhsh, MD, at Johns Hopkins University.

"Sometimes, I'm the third or fourth neurologist a patient has seen," Dr. Nourbakhsh says. "One important element is listening to patient history. Many times we can get clues from what happened to them years ago that can help with the process."

There is no guarantee, however, that the process will move along quickly. "This is a difficult disease to diagnose," Dr. Nourbakhsh says. "Based on the situation, we may be able to tell patients there's a good likelihood of MS, but sometimes we need time."

It's not the best situation for a patient to find themselves in, Dr. Nourbakhsh says. "I could imagine it's difficult for the patient," he says. "The first step would be embracing this uncertainty, being transparent with the patient and letting them know that despite all the testing, talking and examining, I'm still not sure." During that time, those weeks, months or even years, a person can feel hopeless, but there are ways you can continue to move forward, including getting relief for symptoms.

“Uncertainty should not prevent us from treating patients to the extent possible,” Dr. Nourbakhsh says. “For example, whatever this disease is that’s causing nerve pain, there are medications for that pain regardless of the etiology, the cause.”

Dr. Nourbakhsh will even send his patients to other specialists. “Many patients will come to me with prolonged dizziness. They’ve been sent to me because they also have lesions on an MRI. I may not think they have MS, and I don’t know the cause of their dizziness, but I have colleagues who specialize in treating patients with chronic dizziness. I tend to refer patients to them.”

“We shouldn’t forget to provide that treatment while patients are waiting to get more clues about the diagnosis,” Dr. Nourbakhsh says. “Knowing a diagnosis may help prevent future damage, but it won’t help the current symptoms. Treating them should still be a priority.”

Moving forward

Volk, too, decided to start over with a new neurologist. Her neurologist did more tests and eventually diagnosed her with secondary progressive MS. Then he left the hospital, and she had to find a new doctor.

For the next 10 years, Volk was under the care of a neurologist at an MS clinic. But she was never put on a disease-modifying therapy.

“Always, in the back of my mind, I wondered if I really had MS,” Volk says. She thinks her doctor had the same hesitation because, even when Ocrevus was approved for secondary progressive MS, he didn’t prescribe it.

Volk was able to treat some of her symptoms, however.

She found several medications and therapies that have been effective, particularly regular massages.

Then, what started as a simple cold led her down another path, one with more specialists, more tests and more uncertainties.

Complex considerations

Complicating an MS diagnosis even further can be this obvious yet sometimes overlooked reality: “You can have more than one medical condition. You can have MS and something else, like migraines,” says Beverly Layton. She started as a nurse in 1979, and by 1987 she was working with John Whitaker, MD, professor and chair of neurology at the University of Alabama at Birmingham, assisting with research looking for a biomarker for MS disease activity. Now retired, Layton works as an MS nurse consultant.

The possibility of multiple conditions makes it important for people who are being evaluated for a diagnosis to keep track of and make available all their past medical records. “If they’ve seen two or three neurologists already, bring that imaging CD [and] bring those reports,”

Layton says. “If there’s spinal fluid testing, any other neurological testing or laboratory testing, get those results to us so we can sort it out and determine if we need to repeat certain testing.”

“Let’s say a patient has fatigue. Are they having depression?” Layton asks. “Are they having trouble getting up? Let’s say they’re fatigued first thing in the morning. Could they have a sleep disorder? There can be many differential conditions that can cause fatigue, such as anemia or thyroid disorders. Lab testing may be needed to help complete the workup.”

Underlying conditions

Volk had bronchiectasis, a condition in which injury or an infection causes scars and loss of elasticity in the airways that carry air in and out of the lungs. Her pulmonologist suggested it was the result of another underlying condition, common variable immune deficiency, which impaired her immune system, leading to respiratory infections. She is now being treated for that with immunoglobulin replacement therapy (IRT), supplementing her blood with antibodies from donors.

When she next saw her neurologist, he said he’d been following her progress with her other doctors and was hopeful about the new treatment. Volk, however, was nonplussed.

For years, she’d been asking him if he was sure she had MS. “He would say, ‘It has to be MS.’ He’s the only one who supported me this whole time,” Volk says, “and now he says, ‘I don’t think it’s MS after all.’” He told her that she didn’t need a follow up, but since he’d seen her for so long, she could return in a year.

“That really upset me. I felt like I lost the one person who had listened to me. You were the doctor that listened,” she says. “And now you’re not going to listen?”

More follow-ups

Once Knapp made the move to Johns Hopkins, Dr. Nourbakhsh asked her if she had been sick before her sensory problems began. She told him about the cold.

“He said I definitely had transverse myelitis,” she says, which can be caused by a number of conditions, including viral infection or a few conditions that also affect the spinal cord. It can sometimes be the first manifestation of MS.



Illustration by Dave Cutler

“Dr. Nourbakhsh said it might have developed into MS, but we’re not sure,” Knapp says. “He felt more comfortable with not treating me for MS than moving ahead.

“Dr. Nourbakhsh made me comfortable,” Knapp says. “The way he presented the information was very clear, the way he said, ‘This is what I can say definitively, and this is what I cannot say because we don’t know.’”

For now, Knapp says, Dr. Nourbakhsh continues to monitor her, keeping an eye out for new lesions and helping her treat her symptoms.

“For me personally, I’d rather hear, ‘I don’t know,’” Knapp says. “I would love to know ‘yes, you have MS’ or ‘no, you don’t have MS.’ But if they can’t tell me, they can’t tell me.”

For now, they can’t tell her.

“I left my house on a Friday in September at 8 a.m. for a doctor’s appointment. That day, I had a work call at 9 a.m. I thought I’d be home for the call, then all of a sudden I’m in the hospital for three weeks,” Knapp says. “It’s just hard to get your head around that.”

Staying strong

Volk isn’t so concerned with a diagnosis. She just wants to feel better. “I don’t care if it’s MS or not. I really don’t,” she says. “I don’t care if there’s a name to it. I just want to be able to sleep at night.” The uncertainty touches her life beyond the sleepless nights, the pain and the medical tests. “I’m a really strong person,” Volk says, “so I don’t think anybody else gets it.” At first, people understood she was sick, but once she discovered what was wrong, Volk figured she’d be able to fix it, or at least explain to friends and family why she couldn’t do something.

“In the beginning, it was different, but it’s been so long,” Volk says of her 12-year medical

limbo. “I think I’ve lost friends over it. They expect me to do things, and when time after time I couldn’t, they stopped talking to me. I wish they understood.”

Volk is still seeking answers.

“Before I had this, I worked full time, taught at least two classes and was getting a PhD,” she says. “I’m not somebody who’s just going to sit around and feel sorry for myself.”

Volk now teaches as an adjunct professor at Towson University and sits through whole classes if necessary. “I need it for me,” she says. “For my brain. I love to teach.” Continuing to do something for herself, not just for her illness, helps her remain grounded.

Volk remains diligent with her doctors, seeking treatment for her symptoms as best she can. For years, she had counseling, as well, but the cost grew to be prohibitive. Still, she carries on.

“If somebody watched how I get through the day, they would not believe it,” Volk says. “I lie down a lot, but then I gather my energy, and then I get up and do what I have to do.”

Brandie Jefferson is a writer in St. Louis, Missouri. She was diagnosed with MS in 2005.

Learn more about [how MS is diagnosed](#).