

Self-Diagnose Successfully

Patients sometimes understand their baffling conditions better than their physicians. Learn how to build a convincing case and work with your doctor to find answers.

BY RICHARD LALIBERTE

The mystifying symptoms began when Julie Flygare was a law student in Boston. She experienced vivid, terrifying hallucinations while asleep and couldn't move. She felt powerful urges to sleep during the day. Everyday expressions of emotion, such as laughing, made her knees buckle. And the problems seemed to get worse once she started noticing them.

When she mentioned them to her general practitioner, the physician suggested Flygare might have a thyroid problem or depression, or that it was “some strange, rare disorder you'll have to get used to.” It wasn't until Flygare saw a sports med-

icine physician about a running-related knee problem and mentioned the knee buckling that she got a clue. The doctor said she'd heard of a condition that caused symptoms like those and thought it was called cataplexy.

INSTANT RECOGNITION

As soon as she looked up cataplexy, Flygare suspected that was what she had. “What I read was overwhelmingly familiar. But I hadn't put it all together.” Cataplexy is a symptom of narcolepsy, a brain disorder in which boundaries between sleep and waking become distorted and blurred. People

with narcolepsy can lose voluntary muscle control when feeling strong emotions and have excessive daytime sleepiness. They can also experience paralysis while falling asleep or waking up, which can be accompanied by hallucinations.

Her newfound knowledge led her to a narcolepsy specialist at Harvard, who made the official diagnosis and provided her with ways to cope with her condition, including medication, strategic daytime naps, and time management to make the most of the high-energy parts of her day. Her ordeal also provided her with a deep appreciation of how frustrating, isolating, and emotionally taxing it can be to know something is wrong but to have to go through a long and sometimes difficult process to find someone who understands—medically or personally—what is happening. In Flygare's case, it took three years to get from recognizing something was wrong to receiving a correct diagnosis.

PATIENT DETECTIVES

Like many people with unusual symptoms, Flygare's own research helped her diagnose her condition. She looked for and found a narcolepsy expert who agreed she was on to something.

Flygare's experience is not uncommon in the rare diseases community, says Mary Dunkle, vice president of educational initiatives at the National Organization for Rare Disorders. Many people with rare disorders spend years trying to solve mysteries that perplex their doctors. Through experience, these people have learned how to convince clinicians to take them seriously when they think they know what's wrong. We asked for more advice from experts, and from patients who have successfully self-diagnosed.

TRUST WHAT YOU SEE

“There's nothing stopping you from becoming an expert on your condition,” says



RARE DEAL: Julie Flygare experienced mysterious symptoms for three years before her research led to a diagnosis of narcolepsy.



TWO OF A KIND: Priscilla Lopes-Schliep (left), an Olympic runner from Canada, and Jill Viles (right, with her son Martin), a stay-at-home mom from Iowa, both have familial partial lipodystrophy, a rare disease that manifests as overdeveloped muscles in Lopes-Schliep and loss of body fat in Viles.

John Grohol, PsyD, treasurer at the Society for Participatory Medicine, a nonprofit entity that promotes collaboration between patients and health care providers. That's especially true if you have a deep understanding of your own body and brain, says Jill Viles of Gowrie, IA. "We know intimately how we look and how our family looks. No matter how much a doctor interviews and examines a patient, it's no match for a person's ability to recognize distinctive traits."

Viles should know, since a recognition of her family's distinctive features ultimately helped her diagnose her own condition. For almost as long as she could remember, she and her father walked with a waddle and had weakness in their arms and legs. Early in her life, doctors discovered that everyone in her family had high levels of an enzyme associated with muscle damage. These family characteristics led doctors to suspect Viles had a form of muscular dystrophy. Yet she also had contractures—a condition in which normally elastic tissue tightens and restricts movement—in her neck, elbows, and ankles. This unusual combination of features, not evident in anyone else in her family, baffled her doctors and prevented them from making a firm diagnosis.

FOLLOW YOUR INSTINCTS

In college, Viles spent hours in the library poring over books and scientific journals for clues about her condition. Finally, she came across an article about an extremely

rare type of muscular dystrophy called Emery-Dreifuss. In pictures, people with the disorder had distinctive, well-defined arms—a trait that her father shared. Joint contractures were also a feature. But her doctors were unconvinced. "At the time I was first seen, in 1979, many doctors were not familiar with Emery-Dreifuss, which was first described in 1966," Viles says. What's more, the genetic condition is usually passed from mothers to sons, and was initially believed to be due to a sex-linked gene. Later, researchers uncovered rare cases of other inheritance patterns, but at the time the family inheritance pattern made it seem even more unlikely that Viles had it—even to doctors familiar with the disease.

But she persisted and eventually connected with a group in Italy doing research on Emery-Dreifuss. Through genetic testing, the researchers found that she and members of her family, including her father, shared a mutation on a gene known as lamin, or LMNA, which acts as a cellular switch for a variety of functions. "Lamin mutations are associated with dominant inheritance, meaning there are 50-50 odds of the gene passing from a parent with the mutation to a child," Viles says. The mutation's effects are unpredictable, but one potential outcome is Emery-Dreifuss muscular dystrophy. The Italian researchers verified that Viles' self-diagnosis was right.

Yet having Emery-Dreifuss didn't explain all her symptoms. For most of her

life, she'd been rail thin and lacked fat to an extreme degree, especially on her arms and legs. For years, doctors had insisted that this was a manifestation of her muscle disease, but Viles wasn't satisfied. During an internship at a medical lab, she came across yet another exceedingly rare condition, called familial partial lipodystrophy. "It had just recently been mapped to the lamin gene," Viles says—and it was marked by loss of body fat. Again, she was struck by pictures of others with the condition, and recognized aspects of herself or her family in them, such as highly defined muscles and blood vessels. Again, doctors dismissed her suspicions. The prospect of having two almost infinitesimally rare conditions at once just seemed too remote, and Viles didn't pursue it. Instead, she got married and started a family.

MAKE CONNECTIONS

It was Viles' younger sister, Betsy Dopf, who kept partial lipodystrophy on the radar. She, too, had highly defined muscles, especially in her shoulders and biceps, along with mild leg weakness (but not contractures). At a conference organized by a lipodystrophy advocacy group, Dopf became aware of Priscilla Lopes-Schliep, an Olympic runner from Canada whose muscles are so overdeveloped and well-defined she's been accused of using steroids. "When I saw photos of Priscilla relaxing at home, I instantly recognized that her biceps and shoulder development

ANDRE FORGET/CANADIAN OLYMPIC COMMITTEE; COURTESY JILL VILES

“ Sometimes random problems that don't seem related to other symptoms can provide crucial bits of evidence. ”

—MARY DUNKLE

looked just like Betsy's," Viles says. Yet Viles' own arms and legs are spindly, and she uses a wheelchair. Was it possible she shared a genetic condition with a muscle-bound track star?

Viles asked sports journalist David Epstein, author of *The Sports Gene*, to be an intermediary between her and Lopes-Schliep. "We corresponded for a long time about my rationale behind this very unusual request," Viles says. When Epstein finally decided to contact Lopes-Schliep on Viles' behalf, the athlete agreed to cooperate with a researcher and undergo genetic testing. The results confirmed Viles' hunch—both she and Lopes-Schliep had associated mutations on the same gene, which can express themselves in different ways. Fifteen years had passed from the time Viles first suspected she had partial lipodystrophy and an official diagnosis in 2015. Dopf was found to harbor the gene mutation as well. Discovering that a shared defect can produce such dramatically different outcomes has been a major advance in understanding the disorder.

"When you see someone or read something that seems like you, follow that feeling," says Kyle Bryant of Downingtown, PA, who was diagnosed with Friedreich's ataxia, a progressive degenerative movement disorder, after two years of false explanations. "It's not just medical—it's a very internal, personal sense of connection."

DIG INTO THE RESEARCH

The Internet makes self-diagnosis possible in ways that were unimaginable a decade or two ago. "It's a rabbit hole sometimes," Bryant says. "But it might be a fruitful rabbit hole." Not all doctors applaud patients for diving into the confusing and often unreliable world of online information, however. An adage found on social media and coffee mugs says, "Please do not confuse your Google search with my medical degree." But patients say medical schools

spend little time covering unusual disorders, leaving many doctors unprepared to deal with them.

In the absence of answers, keep digging deeper into the medical literature, Flygare and others advise. "Maybe your problem is not this physician's specialty, and you need more investigation."

Flygare recommends casting a broad net to start. "My first step was to look up patient organizations for narcolepsy and read their background information." She also set up news alerts to automatically scan the web for selected keywords and deliver bundles of new hits at specified intervals. "They're not all relevant," she says. A well-known keyword like "narcolepsy" can snag useless references such as jokes. "But 'cataplexy'—nobody has heard of it outside a certain community, so those searches catch just the important things," she says.

If you're not sure what keywords to use, start with symptoms—and don't write off minor tics that seem inconsequential. "Sometimes random problems can provide crucial bits of evidence," Dunkle says. Take notes about everything that occurs and when, she advises.

SEPARATE GOOD RESEARCH FROM BAD

Confirming a diagnosis requires insight about the problem beyond a hunch or hearsay off the Internet, especially when the cause isn't clear. "One mistake people make is latching onto information they find interesting but doctors wouldn't put their faith in," Viles says. "You really need to learn the language doctors speak. Anchor your thoughts to published medical citations. That's something a doctor would look for."

Some medical sources are more credible than others. Journals should have a peer-review process in which independent experts validate that a study's findings are legitimate, says Dr. Grohol. Name recognition helps. "Doctors will probably accept

evidence from a known journal more than one they've never heard of," he says.

Not sure how influential a journal is? Check the publication's website for what's called its impact factor. "That's a measure of how many other researchers are citing it as part of their own research because it's important," Dr. Grohol says. "You're looking for an impact factor greater than 1 or 2." For example, *The New England Journal of Medicine*, a renowned publication covering a variety of subjects, rates 55.9. *Neurology*, the leading scientific journal in the field of neurology, rates 8.166. "But probably half of the tens of thousands of journals out there rate lower than 1," says Dr. Grohol.

Find leads on more credible sources or experts by looking at medical or scientific advisory boards of organizations that focus on certain conditions. Search for experts giving keynote addresses at professional, medical, and scientific conferences. Or look at general interest publications to see which experts are often quoted. Flygare identified the nation's best specialists and treatment centers for her condition in articles in *The New York Times*. "Look at what the leaders are talking about and putting their names on," she says.

FIND A COMMUNITY

People experiencing your symptoms or a suspected condition may understand the situation in ways doctors don't. "It's helpful to seek a patient community where you can discuss findings with other people," Dr. Grohol says. "You can ask questions like, 'Am I crazy and going down the wrong road?' Having a reality check and feedback can help you understand possible next steps."

Connecting with a community can also advance science. That's what happened when Matthew Might, PhD, associate professor of computer science at the University of Utah, wrote about his son's movement disorder after it was identified



MIGHT MAKES RIGHT: Matthew and Cristina Might with their children Victoria, Winston, and Bertrand, who has a rare movement disorder identified as a double mutation on a gene called NGLY1.

as a double mutation on a gene called NGLY1. With the hope of finding more patients with the mutation—his son was the only person in the world known to have the defect—Dr. Might wrote a provocative blog post designed to go viral. First came a grabby headline: “Hunting Down My Son’s Killer.” Then the words: “I found my son’s killer. It took three years. But we did it. I should clarify one point. My son is very much alive. Yet, my wife and I have been found responsible for his death.” Several thousand words of medical details and a link to the research followed. As he hoped, the post took off on other blogs and social media outlets like Twitter, and within three years, 35 more cases of the disorder were found worldwide. (Read the post at bit.ly/MySonsKiller.)

Now an advisor to the Undiagnosed Diseases Network at the National Human Genome Research Institute, Dr. Might says patients are shifting the way rare diseases are discovered, studied, and treated in part by revealing and sharing new information—an approach pioneered by real-life families portrayed in the movies *Lorenzo’s Oil* and *Extraordinary Measures*.

For the majority of people who have symptoms or conditions with larger patient populations, blogs and social media platforms like Twitter and Facebook can

help you find fellow travelers. “Look for Twitter hashtags like #raredisease and follow those feeds,” Bryant suggests. If you think you’re dealing with a particular disorder, you may find related groups on Facebook. “Groups are a mixed bag,” Flygare says. “Read posts with a grain of salt and know that everyone has different experiences. But they’re a good way to get more information.”

WORK WITH YOUR DOCTOR

Those who diagnose themselves can’t go it alone. They need confirmation, guidance, and treatment from doctors. Patients may worry that doctors won’t agree with their self-diagnosis and that if they don’t, their doctors won’t help identify the real problem. But the doctor-patient relationship shouldn’t be viewed as a battle, says Dunkle. “It’s really important to have a good relationship with your doctor and keep the search for a diagnosis positive.”

Cull your research. One way to smooth the relationship is to determine what’s important to talk about during a doctor visit. That includes sharing only relevant research. For example, Viles brought her doctor literature on lipodystrophy and photographic evidence of the condition when she began re-investigating it. It prompted Viles’ physician to read exten-

sively on lipodystrophy, which ultimately convinced him she might have it. But not all patients are as focused. “I’ve had patients ask me to look through hundreds of pages of information they found online,” says Bruce H. Cohen, MD, FAAN, director of the NeuroDevelopmental Science Center at Akron Children’s Hospital in Ohio. “There aren’t enough physicians in the world to go through all the data some patients bring.”

Each case is different, and “ideally, the doctor works with the patient to highlight and focus on the kinds of things they think might be most relevant,” says Dr. Grohol. When sharing research of your own, Dr. Grohol suggests concentrating on findings you think are most important or specifically pertinent to your condition.

Prepare three questions. Focus and brevity are key, says Flygare. “If you have 20 questions, ask a friend to help you narrow them down,” she says. If you’re at a loss for what to ask or seem to be at a dead end, Flygare suggests asking “What other direction, specialist, or expert should I consider? How should I proceed from here? What resources or patient groups are available for me to learn more?” Keep your time with the physician focused on your medical issues. “You’re not looking for the doctor to hold your hand or give you a shoulder to cry

“ You have to find people who work for you and help you make progress. It’s **important to be persistent**. Keep going until you find the answers you need. ”

—KYLE BRYANT

on,” she says. “You can get that from other people. You need the best medical care, and you need to schedule your time and your doctor’s time around that.”

Define symptoms. Dr. Cohen urges patients to build on their medical history by shifting from describing symptoms to telling your doctor about signs of disease that other physicians may have identified by name and relayed to you. A *symptom* is something you alone experience, such as pain or headache. A *sign* is something that someone else

can objectively observe. For example, he explains, rather than saying you have blurry vision, which is a symptom, you would tell a doctor you have optic atrophy, or deterioration of the optic nerve, which can cause blurry vision. A sign isn’t the same as a diagnosis, Dr. Cohen says, but it narrows a symptom down to a specific clinical concern that can help clarify the problem for everyone in the diagnostic chain. “That can help physicians and patients piece together the puzzle leading to diagnosis of the underlying trouble,” he

says. Optic atrophy, for example, can occur with multiple sclerosis.

Don’t antagonize. Keep the tone collegial and friendly, says Dr. Grohol. “Both you and your doctor have information to bring to the conversation. Your job is to raise yours in a way that’s respectful and makes a point without putting down the medical establishment,” he says. And don’t hold lack of knowledge against a clinician, Flygare says. “I love when a doctor says to me, ‘I don’t know.’ I prefer that to misinformation or educated guesses,” she says.

Available Resources

- ▶ **PUBMED:** Run by the National Institutes of Health (NIH)’s National Library of Medicine, this searchable database is an indispensable resource when searching for scientific studies in the medical literature. pubmed.com
- ▶ **UNDIAGNOSED DISEASES NETWORK:** This clinical research initiative from the National Human Genome Research Institute recently added six sites around the country where researchers from different disciplines collaborate to speed diagnosis of baffling genetic disorders. An online portal launched in 2015 allows the public to apply directly to the NIH for case review. bit.ly/UDNInfo
- ▶ **OMIM:** The Online Mendelian Inheritance in Man catalog from Johns Hopkins University provides a way to identify possible genetic disorders by typing symptoms into a search field. It’s meant for doctors and other professionals but is open to the public. omim.org
- ▶ **NORD:** The National Organization for Rare Disorders vets organizations that offer high-quality information about specific conditions and provides links to their websites. A Rare Disease Information database offers A-to-Z descriptions of more than 1,200 disorders. rarediseases.org
- ▶ **GARD:** The Genetic and Rare Diseases Information Center from the NIH provides searchable overviews of an extensive range of conditions, with links to further information. Users can ask specialists questions through an online contact form or toll-free number. bit.ly/NIH-GARD
- ▶ **CITEFACTOR:** This searchable database lists impact factors for academic journals, providing an indication of how often researchers cite these publications’ studies. citefactor.org
- ▶ **WIKIPEDIA:** Written by users, this crowdsourced encyclopedia “is only a start and is sometimes written by people with agendas, but it can raise helpful questions for you to ask your doctor,” says neurologist Bruce H. Cohen, MD, FAAN, of Akron Children’s Hospital. And footnotes can lead you to primary sources. wikipedia.org

DON’T GIVE UP

“If a physician is reluctant to work with you as a team, that’s probably not a good doctor to work with in the long term,” says Bryant. “You could literally be fighting for your life, and you don’t want to be fighting both your disease and the person who’s supposed to be helping you.”

If you’re not getting answers, don’t be afraid to get opinions from different physicians. “A lot of people get stuck in a rut thinking that if they go to another doctor, it will offend the first doctor,” Dr. Grohol says. “But it’s important to get second opinions about a serious illness and not just accept one person’s view.” When reaching a roadblock, Flygare says it’s helpful to ask, “What needs to happen to resolve this?”

In the end, Flygare’s persistence led to treatment and a semblance of a normal life. “Adjusting to medications and lifestyle changes took a few years, and I’m still learning,” she says. “But I’m doing well, working full time, running a nonprofit organization called Project Sleep in my free time, and living a full life.”

“You have to find people who work for you and help you make progress,” Bryant says. “It might be five or six people down the road, but it’s important to be persistent. Keep going until you find the answers you need.” NN