Medical Mystery: When You Can’t Get a Diagnosis

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Her symptoms came on slowly but kept getting worse. Within a few months, Susan Warm’s fatigue had turned into debilitating exhaustion, her aches had graduated to serious pain, and her periodic sweats had developed into waterfalls that plastered her hair to her head. Warm, then 45, consulted several doctors, each of whom ran a variety of tests and declared she was suffering from nothing worse than the symptoms of menopause. “I thought I was going crazy, because I felt so bad for no explainable reason,” recalls Warm, a customer-marketing specialist in Pasadena, California.

Eight months into her ordeal, Warm persuaded an ER doctor to admit her to the hospital. There, one of the attending physicians touched her drenched arm and said, “I haven’t felt sweats like this since I last treated tuberculosis.” Sure enough, even though Warm didn’t have the classic TB cough, she tested positive for the disease. (The infection was in her bloodstream, not her lungs.) Once Warm took the appropriate antibiotics, she was cured—at last.

Americans believe that diagnosis is the cornerstone of medical care: You go to a doctor, who labels your problem and prescribes a treatment that makes you well. But what if the process gets stuck at the labeling stage? “We have a fantasy that as soon as we describe our symptoms, the doctor will know what is wrong with us. But the reality can be much more complicated,” says Evan Falchuk, president of Best Doctors (bestdoctors.com), a Boston-based company that helps corporate employees get second opinions from top physicians.

You don’t expect the doctor to be stymied, particularly after extensive testing. But failures to find a diagnosis do occur—and while no one knows exactly how often, 10,000 patients contact Falchuk’s firm for help every year.

TV shows like House contribute to our misconceptions. Although Dr. Gregory House is portrayed as an expert in diagnostic medicine, such a medical specialty exists only in scriptwriters’ imaginations. In real life, analyzing what’s wrong with us falls to our primary care physician or to the specialists we consult. In most cases, by relying on our symptoms, medical history and test results, doctors are able to figure out our problems. But MDs typically look for common scenarios; people with rare diseases or unusual presentations of prevalent conditions (like Warm’s TB) can fall through the cracks, notes Marianne Genetti, executive director of In Need of Diagnosis (inod.org), a nonprofit in Orlando, Florida, that assists patients who have not been able to put a name to their complaints.

If that’s your situation, don’t give up: It’s essential that you persistently advocate for yourself. One way to do that is to actively help your doctors find the correct diagnosis. Here, five steps you can take.

**Step 1: Do Your Homework.** The more information your doctor has, the better he’ll be able to treat you. In the days leading up to an important doctor visit, keep a pad of paper handy and jot down your symptoms as they occur so you can read the list to your practitioner. In addition, do a little digging on the Web, spending time on reputable medical sites...
such as MayoClinic.com and Health.gov to gain insight into how your symptoms might fit together. For instance, you may have gotten so accustomed to feeling thirsty all the time that you didn’t even think to bring it up during your last visit. But if an online check reveals that the symptoms you did plan to mention—fatigue and headaches—often go along with increased thirst in diabetics, then you’ll realize you should be telling the doctor how frequently you need a drink of water.

Or maybe you have a funny bump on your hand. By going online, you discover that one possible cause is a marine parasite. Now you remember that you swam in the ocean during your last Caribbean vacation, so you’ll be sure to tell the doctor.

Just don’t try to generate your own diagnoses, cautions Marie Savard, MD, an ABC News medical contributor and author of How to Save Your Own Life: The Eight Steps Only You Can Take to Manage and Control Your Health Care. Online information helps you pose smarter questions to your doctor, but he’s much better qualified to piece together the information.

At your doctor’s appointment, you’ll also need to list all the medications (and supplements) that you take or have recently taken. Unexplained symptoms frequently stem from a reaction to a drug, even one you’ve been on for a while, says Abbie Leibowitz, MD, chief medical officer of Health Advocate (healthadvocate.com), a firm in Plymouth Meeting, Pennsylvania, that helps patients navigate the health care waters.

But the best information you can give a physician is your personal and family medical histories. “Up to 80 percent of a doctor’s determination comes from your history, not from tests done the day you’re in the office,” Savard says. Be ready to describe all the major conditions you have ever had, even if they are long in the past or do not seem associated with your current symptoms. Bring to your appointment important test results from any related prior illnesses.

And make sure to tell your physician about conditions your relatives have suffered from. Rare diseases often go undiagnosed, but if your doctor knows that one runs in your family, he will be much more likely to consider it a possibility.

**Step 2: Prod Your Doctor.** When symptoms don’t add up to a ready diagnosis, physicians sometimes suggest that an ailment is psychological or stress related. But don’t let your doctor dismiss your complaints so quickly, says Pamela Hops, MD, a family physician at New York City’s Continuum Center for Health and Healing. “Sometimes psychological issues do present themselves physically, but the doctor shouldn’t assume that’s the case, especially if you feel in your gut that something else is wrong,” she says. Some symptoms, says Hops, can indicate a serious condition and always call for additional testing: prolonged fever, unexplained weight loss and night sweats that aren’t associated with menopause. Night sweats, for example, can indicate an infection or a possible cancer or may be a side effect of certain diabetes drugs.

If your doctor is baffled, suggest he consider conditions that are known for being overlooked. (See 5 Diseases Doctors Often Miss) Take, for instance, Lyme disease, which sometimes surfaces in the form of vague and ordinary symptoms. Stephanie Smith, 36, an asset manager from Newport Beach, California, suffered for a decade from joint pain, fatigue, insomnia and heart palpitations before she was diagnosed with the disease, which is primarily a problem in the northeastern U.S., not in her home state. When one doctor suggested her troubles were due to stress, Smith (who asked that her real last name not be used) quit her high-pressure job, but she continued to deteriorate. “I began to think my symptoms were normal age-related issues, and eventually I stopped trying to get a diagnosis,” Smith says. Then one day she described her condition to her aunt, who’d once had Lyme disease and recognized the symptoms. After a positive test, Smith started using the antibiotics and immune-boosting supplements that returned her to health.

**Step 3: Take a Break.** If your doctor has run important tests and ruled out serious conditions but still hasn’t determined what’s wrong with you, it may be time to put your quest for a diagnosis on hold. “You don’t want to push your doctor into giving you drugs or other treatments that may not be appropriate, because that can lead to other complications,” Savard says. She suggests waiting for three months, or whatever time frame you and your physician agree on, to see if your body can heal itself.
During this hiatus, you might take a more holistic approach to healing by adopting a healthier lifestyle: Get enough sleep, exercise regularly, reduce your stress levels and improve your diet—and see if these measures are enough to cure what ails you.

Changing your diet can be tough, but in some cases the results are dramatic. For years, Kristen Johnson, 51, who lives in San Diego, suffered from migraines, digestive problems, depression, aches and pains. Then she decided to seek out an MD with a holistic bent (find doctors with this orientation through the Institute for Functional Medicine, functionalmedicine.org). The physician thought she might be severely sensitive to certain foods, especially wheat, corn, soybeans and sugar. Eager to try anything that could help, Johnson made over her pantry. “I swooped into my kitchen, tossed out all the processed foods and stocked up on vegetables, fish, poultry, fruits and yogurt,” Johnson recalls. After a couple of weeks, she found relief, and within a few months, she felt truly healthy. “I still get the occasional migraine, but it’s always after I accidentally eat something hidden in restaurant food,” explains Johnson.

Step 4: Call in the Experts. If you’ve followed the advice above and haven’t made progress, it’s time to seek out a topflight physician in a specialty related to your concerns (for instance, a neurologist if you have unexplained tremors). “Always keep your regular doctor in the loop, because if you still don’t have answers after seeing the expert, you’ll want to return to your doctor to discuss other options,” says Leibowitz of Health Advocate.

The best specialists can be found at large hospitals or, better yet, at teaching hospitals affiliated with medical schools—even if these are a long drive away. It’s especially important to seek this top level of expertise once you’ve seen and stumped other doctors, since that means you’re more likely to have a rare condition, says William A. Gahl, MD, PhD, director of the NIH Undiagnosed Diseases Program.

“Doctors at a university center usually have the most up-to-date information on rare diseases, and they have an array of specialists that can address them,” he says. Working through your physician, you can also get second opinions without an in-person examination from such top-rated institutions as the Cleveland Clinic (clevelandclinic.org; click on My Consult), Johns Hopkins Medicine (hopkins-medicine.org/second_opinion) and Partners Online Specialty Consultations (econsults.partners.org), which connects to Harvard-affiliated doctors. The fees might be hefty but could be worth the expense.

Step 5: Be Prepared for a Diagnosis to be Wrong. Sometimes the problem isn’t that you’re unable to get a diagnosis but that you’ve received an inaccurate one. Mistakes are not uncommon: Autopsies performed on patients in one university hospital found that up to 32 percent of them had been given serious misdiagnoses. “If your treatment isn’t making you feel better, don’t immediately look for other therapies. Your first move should be to confirm that your doctor got the diagnosis right in the first place,” says Falchuk of Best Doctors. That typically involves sending all your records to an expert or, in the case of cancer, asking that a different pathologist examine your tumor sample. One of Falchuk’s clients, who was being treated for cervical cancer, discovered in this way that she actually had colon cancer that had spread to her cervix. Because cancer treatment is specialized, she had been getting the wrong drug. “It’s important to keep asking questions to ensure your diagnosis is right,” Falchuk says. “The worst that can happen is that your doctor may feel annoyed.” And what’s the best that can happen? Your questioning may well save your life.