https://www.washingtonpost.com/national/health-science/at-first-a-man-thought-he-had-food-poisoning-not-so/2015/10/26/71290448-4e61-11e5-902f-39e9219e574b_story.html?utm_term=.7a7d354131e2

The Washington Post

At first, this man thought he had food poisoning. It turned out to be something far worse.

By Sandra G. Boodman **Reporter** **October 26, 2015**

(Robert Carter for The Washington Post)

Jeffrey Sank always knew when he was going to have an attack: Its onset was signaled by the same kind of uneasy, “uh-oh” feeling that portends an impending cold.

But Sank’s problem wasn’t in his head — it was in his gut. And when he felt the initial abdominal pangs, he knew that he had about 12 hours before he was miserable, or at worst incapacitated, for the next day or two.

“It almost felt like I’d done 1,000 sit-ups or been punched in the gut 100 times,” said the digital media specialist, 43, who lives in the District. At first the attacks were intermittent. But after several months

the pain, centered in the right upper quadrant where the liver and gallbladder are located, increased in severity and frequency. *[*

For nearly a year, Sank, with the help of his stepmother, a physician, struggled to determine the reason for his pain. He saw multiple doctors, including two gastroenterologists, a kidney specialist and an infectious-disease physician. He underwent workups for reflux disease, a liver disorder, an intestinal blockage and malaria. One doctor suspected he might be faking.

Sank's problem turned out to be none of those things. His diagnosis was partly the result of serendipity: The second gastroenterologist he consulted was familiar with the malady, which is common in other parts of the world but not the United States. To complicate matters, Sank's case did not fit the standard diagnostic criteria.

"It's in the differential [a list of possible disorders suggested by symptoms], but since we never really see it, you don't necessarily think of it," said Montgomery County gastroenterologist William Steinberg.

Luckily, something Steinberg had seen two decades earlier on a medical trip to the Middle East resonated when an increasingly desperate Sank consulted him in April 2010.

The first time he suffered stomach pain in June 2009, Sank assumed he had food poisoning. "I really didn't think much about it," he recalled.

When it kept recurring, he consulted his stepmother, Catherine Shaer, a retired pediatrician, for advice. Sank was otherwise healthy, and Shaer agreed that he should see a gastroenterologist.

At his initial appointment in October 2009, the gastroenterologist told Sank he suspected his pain was the result of gallstones and ordered a sonogram.

Prepare yourself, doctor warned

The test was memorable: Sank said that during the procedure the technician began acting strangely and then summoned a radiologist. In a somber voice, Sank recalled, the radiologist "told me that there was a huge lesion on my liver and they were going to send me immediately for a CT scan."

Jeffrey Sank with his stepmother, Catherine Shaer, a retired pediatirician whose advice helped lead to the proper diagnosis of his medical problem. (Family Photo)

The radiologist then told him, Sank recalled, “there was something very serious going on here and that I needed to prepare myself and my family for what I had to deal with.” Sank also remembers the specialist saying that his “door was always open.”

“I thought I had liver cancer and was going to die,” Sank remembered. At the time, his first child was only a few months old.

While waiting for the CT scan, Sank telephoned his stepmother and a friend who is an oncologist. Both told him that they were sure that the growth on his liver, the size of a large strawberry, would turn out to be a benign hemangioma. He had no cancer symptoms, and such tumors are common. A few hours later, Sank, hugely relieved, learned they were right. He didn’t have cancer. Nor did he have gallstones. “We were back to square one,” he recalled.

His oncologist friend suggested that during the next attack, Sank go to an ER and ask for a CT scan, which might reveal something about what was wrong as it was occurring. Sank had also discovered that during the episodes he ran a low-grade fever — less than 101 degrees — which suggested a possible infection.

In December 2009, in the throes of an attack while at work, Sank took a cab to a Washington emergency room.

The staff, he said, was a “bit dismissive.” One resident suspected he might be malingering, according to his stepmother, who reviewed his medical records.

A CT scan showed nothing abnormal, but blood tests did. His level of C-reactive protein, which measures inflammation, was sky-high at 148 milligrams per liter. A CRP reading above 3 would indicate a high risk of cardiovascular disease. Something was clearly causing an inflammatory response, but doctors couldn't tell where the problem was, or what it was. Sank was sent home and within a day or so he was back to normal — until the next attack.

Down the rabbit hole

His gastroenterologist, Sank recalled, seemed stumped and disinterested. "He was still talking about taking out the gallbladder, just in case that was the problem, which didn't make any sense to me."

At the suggestion of another friend, Sank decided to consult a Northern Virginia chiropractor. "I figured, 'What the hell, maybe he could figure it out.' " The chiropractor ordered numerous tests and took stool samples. He told Sank that he had "second-degree liver failure" and prescribed expensive supplements designed to correct the problem.

Shaer, who considered the chiropractor a quack because of his unscientific approach and dubious treatment, said she "freaked out." At her urging, Sank agreed to dump the chiropractor and see her internist instead.

The internist was concerned that sporadic fevers might signal an infection, possibly malaria contracted during trips abroad; Sank had spent time in Thailand and Mexico five years earlier. He referred Sank to an infectious-disease specialist.

Subsequent tests ruled out malaria as well as porphyria, an inherited disorder that can cause abdominal pain. Because Sank had undergone an appendectomy in 2004, doctors next wondered whether he had developed adhesions, a form of scar tissue that can cause recurrent pain. That, too, proved to be a dead end.

In March, Sank noticed blood in his urine and consulted a kidney specialist. "He did tons more blood and urine tests," Sank said, but nothing significant emerged.

"By this point I was pretty confident I didn't have something horrible, or they would have found it," he added. But the attacks, which were occurring on a weekly basis, were draining and disruptive. "I was concerned about how long I was going to have to deal with this."

His stepmother persuaded him to see Steinberg, her gastroenterologist.

Out of the past

During Sank's first appointment, Steinberg said he was struck by the extremely high CRP levels, which invariably returned to normal after an attack.

"I'm thinking, 'What could do that?' " Steinberg remembered. Sank had already undergone three CT scans and a plethora of blood tests, which had ruled out the usual suspects: pancreatitis, a perforated ulcer and a rare hereditary disorder called angioedema.

Steinberg said he wondered about an illness that did not seem to fit Sank's case.

Twenty years earlier, the gastroenterologist, who has practiced in Ethiopia and Indonesia, traveled to Israel on a medical trip. While there, he had discussed with local doctors a common inherited disorder seen frequently in Israel, Turkey and Greece known as familial Mediterranean fever (FMF). One Tel Aviv clinic Steinberg toured was following 2,500 patients with FMF.

The disorder is caused by a mutation in the MEFV gene, which provides instructions for making a protein found in white blood cells; the first episode usually occurs in childhood or young adulthood. It is characterized by a red rash on the lower legs, achy joints and abdominal pain. Sank was 38 and had no history of a rash or achy joints. Nor, as far as he knew, did anyone in his family, including his older brother. He was descended from Eastern European, or Ashkenazi, Jews who settled in Austria. FMF is known to affect Sephardic Jews, whose ancestry is Middle Eastern, as well as non-Jews from the Middle East, Italy and Armenia. In most cases, those who inherit one mutated gene are carriers; those with two copies of the gene, one from each parent, develop the disease.

If FMF is not treated, it can cause kidney failure, arthritis and a potentially lethal disorder called amyloidosis. Although there is no cure, treatment involves medicines that reduce inflammation and prevent attacks. In about half of people with FMF, episodes are preceded by milder symptoms called a prodrome, that feeling of impending illness Sank repeatedly experienced.

Although Sank said he knew of no Middle Eastern connection, Steinberg was unconvinced. "You never know what's floating around in our blood," he said. It turns out that he was not the first doctor to wonder about the diagnosis. Records show that the infectious-disease specialist considered FMF but did not pursue it after Sank told him he was of European descent.

Steinberg thought it might be worth giving Sank a course of colchicine, a well-tolerated drug most commonly used to treat gout. Colchicine is also a mainstay therapy for FMF; it reduces inflammation and prevents attacks.

He conferred with another doctor in his practice, NIH-trained rheumatologist Rachel Glaser, who has significant experience treating FMF patients; she agreed it was worth a try.

The drug had a dramatic effect. After Sank began taking it, his attacks largely ceased.

Several months later, at the suggestion of Glaser, who had become his treating physician, Sank underwent a genetic test that confirmed the diagnosis of FMF and revealed that he had inherited mutated copies of the affected gene from both parents.

"This had thrown my life into a tailspin," Sank said, "so I thought it was good news, even though it meant that I had a chronic disease." He continues to take colchicine and sees Glaser and a kidney specialist once or twice a year.

It is too early to tell whether his two young children have inherited the flawed gene. If they develop symptoms, they will be tested, he said.

For Sank, having a stepmother who is a doctor has been a big help. Shaer credits Steinberg's willingness to think unconventionally.

Steinberg said the memory of what he learned during his travels in Israel proved to be crucial two decades later. "It's just very uncommon in America," he said of the disease. During a career that spans more than 40 years, Sank's remains the only case he has treated.