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Medical Mystery: Alcoholism didn't cause man's diabetes and cirrhosis

By Sandra G. Boodman, Published: June 13

When the medical resident repeated what other doctors had insisted was the cause of her twin brother's sudden, serious illness, Janet Janas lost it.

“I think *you* need to rethink your judgment because that has *not* been established,” the normally unflappable pediatric nurse practitioner recalls saying. The resident, she said, rolled his eyes, telegraphing his frustration with a relative intent on denying the obvious.

Doctors had told Janas and her family, who had gathered in the intensive care unit of a Portland, Ore., hospital in January 2008, that her brother Jeff Williams, delirious and combative, was in the throes of advanced alcoholism. Doctors told the family he had [cirrhosis](#) of the liver, internal bleeding and [ketoacidosis](#) — a medical emergency in which a shortage of insulin can lead to coma or death — from newly diagnosed diabetes.

When Williams's family protested that they considered the 46-year-old electronics engineer, who had been in good health, to be strictly a social drinker, the staff disagreed. “They can hide it really well,” Janas said one nurse told her.

Williams said he confronted similar disbelief during and after his two-week hospitalization. “I said, ‘I drink about one beer a day, not enough to cause cirrhosis, so this just ain't the cause,’ ” he recalled telling doctors repeatedly. “They just kind of blew me off and said, ‘Well, it is what it is.’ ”

Ultimately it was Williams's ferocious determination to figure out what was wrong that led to a diagnosis that affected his entire family, especially his twin sister.

Worsening eyesight

In November 2007, Williams spent a week visiting Janas at her Northern Virginia home. She said her normally intense brother seemed “mellow” than usual, even napping every afternoon, which she attributed to his relaxing on vacation.

Back in Portland a few weeks later, he suddenly developed blurry vision. Janas told him to call his doctor; his HMO referred him to an optometrist, who gave him his first pair of glasses.

Within days his vision had gotten worse. Janas urged her brother to call his doctor; instead, he was sent back to the same optometrist. He asked why his vision had deteriorated so quickly. "That can happen," she replied, before giving him a stronger prescription, denying his request for a referral to an ophthalmologist.

A few weeks later, Williams thought he had contracted a bad case of flu; he was exhausted and achy. On Dec. 28, he had trouble getting out of bed and, severely dehydrated, "drank gallons of Gatorade," and went to see a doctor at his HMO's urgent care center. His blood sugar was alarmingly high at [371 milligrams per deciliter](#) — nearly double the threshold that triggers a diabetes diagnosis. The doctor sent him home with diabetes medication.

Williams said he doesn't remember coming home but managed to call his 20-year-old daughter, who lives nearby. "I said, 'I'm in bad shape, and if I don't get better, take me to the hospital,'" he told her. The next day, she discovered him disoriented and barely conscious, and rushed him to the hospital. His blood sugar was above 500, his abdomen was swollen with fluid, his spleen was enlarged and he was vomiting material that looked like coffee grounds, signaling internal bleeding.

"They told me had I come in 12 hours later I would be dead," said Williams, who was dispatched to the intensive care unit.

Looking for the cause of these symptoms in a thin, previously healthy, athletic man, doctors ruled out infection, hepatitis C and other diseases, and settled on what they deemed most likely: [alcohol abuse](#). His delirium and confusion were regarded as signs of withdrawal, and the ketoacidosis was a complication of [Type 1 diabetes](#), an unusual diagnosis in a 46-year-old, because most cases occur in children and young adults.

Hospital records show that Williams strongly denied a drinking problem but that he "freely acknowledges considerable alcohol abuse" in his youth. His brother and daughter said they didn't know how much he drank but told hospital staff there was a beer dispenser known as a kegerator in the basement of the home he shared with roommates. Williams's father adamantly denied that his son was an alcoholic; Janas was similarly incredulous.

"We're a very close family," she said, adding that the idea that her twin brother could have harbored such a secret for years seemed unfathomable. The week he spent at her house, he'd drunk only an occasional beer.

When she told one nurse about her skepticism, Janas said, the response was frosty condescension, an attitude that Williams said he also encountered. "I hadn't even had a drink for the two weeks before I was hospitalized, because I hadn't felt well," he said. "So how could I be in withdrawal? It just made no sense."

'That's what I have!'

Over the next few months, Williams slowly recovered, adjusting to life as an insulin-dependent diabetic with a badly damaged liver. Determined to find the cause of his health problems, he called his longtime friend Delbert Scott, a retired pathologist. In May, he spent several days at Scott's ranch in eastern Oregon.

“We were discussing his history and diabetes, and for some reason we got out my Merck Manual and looked up the possible causes of liver failure, and there it was,” Scott recalled. One of the causes of cirrhosis and diabetes is [hereditary hemochromatosis](#), a genetic disease that causes the body to absorb and store too much iron, causing organ damage.

“I read it and said, ‘Oh my God, that’s what I have!’ ” Williams remembers saying. Scott was not convinced: His friend lacked the telltale copper-colored skin. “The people I’d seen looked pretty bronzed,” Scott said.

One of the most common genetic diseases in the United States, hereditary hemochromatosis affects an estimated 1 million Americans, most of them, like Williams, Caucasians of Northern European descent. About five of every 1,000 Americans who inherit two copies of a defect in the iron-regulating [HFE gene](#) discovered in 1996, are most at risk, according to the National Institutes of Health, but not all of them will develop hemochromatosis. Those who inherit a defective gene from only one parent — about 10 percent of the population — are carriers; they can unknowingly pass the gene to their children but are less likely to develop problems than people who inherit two copies of a defective gene.

In its later stages, hemochromatosis can cause cirrhosis and diabetes as well as heart failure. It is detected through blood tests, including one that measures serum ferritin, which, if elevated, may trigger a genetic test.

Unlike most genetic diseases, there is effective treatment, and it is simple: regular blood draws of about a pint or so to reduce iron levels, followed by maintenance phlebotomy to keep the iron level in check.

Williams said he immediately called his HMO and demanded the relevant tests. His ferritin level was 2,350, far above the 300 nanograms per milliliter generally considered the upper range of normal for men. He soon learned that he had inherited two copies of the gene and that his supposition was correct. Hemochromatosis, not alcohol abuse, explained the cascade of diseases that landed him in the hospital: cirrhosis, diabetes heralded by the abrupt vision changes, even the apparent case of the flu, which was actually ketoacidosis.

“I was just so pissed,” said Williams, who immediately began weekly phlebotomy sessions. Janas quickly discovered that, like her twin, she had inherited two copies of the gene and was in the early stages of the disease; she immediately began weekly blood draws and after eight months her ferritin reading dropped to a safe level. Their older brother learned he is a carrier, as is Janas’s teenage son; both are being monitored.

“She’s done well,” said Lee Resta, a Northern Virginia hematologist who has treated Janas since her diagnosis.

In May 2009, Williams obtained his hospital records. Buried in his discharge summary was something that left him reeling. Doctors had, in fact, tested his serum ferritin level and found it “markedly elevated” at 2,098 nanograms. They never ordered a genetic test because they thought the level was the consequence, not the cause, of his cirrhosis.

Resta said he finds the decision not to pursue the abnormal finding baffling. “With liver disease and a ferritin level of over 2,000,” he said, “why not check it?”

Williams said he has never received an explanation he considers satisfactory. He said he decided against filing a malpractice suit because there was no way to prove that the misdiagnosis caused harm; his liver damage had probably occurred well before his hospitalization.

He has tried to channel his anger into raising awareness of the disease and advocating for screening; several people he works with discovered they had the gene as a result of his experience, he said. Williams said he is scrupulous about taking care of himself, and faithfully injects his insulin and tests his blood sugar four times a day to control his diabetes. The specter of liver cancer, a consequence of irreversible cirrhosis, hangs over his head. It took nearly 18 months of weekly blood draws to reduce his iron to a safe level.

“The only reason I’m doing well is that he’s not,” observed Janas, who said she feels both guilty and grateful. “Without him, I’d just have gone on my merry way not knowing. But the pain that he went through, that my family went through . . .” she added, her voice trailing off.

“She’s the case of what happens when things are done correctly, and I’m the case of what’s done incorrectly,” Williams said. He remains furious that doctors wrote him off as an alcoholic in denial and wonders what might have happened if he hadn’t made the discovery that led to the truth. “It was the engineer in me that just wouldn’t let this go.”

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