

Memoir | *Ludwig Gutmann, MD*

Never saw it coming

Fred never was very strong, so it was not clear when his weakness began. He was in his early forties when I first saw him, many years ago.

As he sat on the exam table in his underwear that first day, loosely covered by a white sheet, he looked like a man who had avoided strenuous activities all his life. His face, arms, and legs were thin and spindly, belying the fact that he had worked in the construction trade for years. The give-away was his coloring—the exposed parts of his body were nut brown from years of labor in the summer sun, and the pale and pasty appearance of his chest and upper arms showed the clear outline of his perennial T-shirt. His lackluster physique seemed to mirror his personality and the nature of his complaint.

His chest musculature was sparse, although his breasts were surprisingly large. His abdomen pouched out over his shorts. He had the resigned, stoop-shouldered look of someone who knows good things never happen.

“I’ve been a carpenter ever since I got out of high school, but over the last few years I’ve been having trouble hanging on to nails and my tools—they seem to slip out of my fingers. My wife says my speech is a little slurred, too. Everything just seems to be getting a little worse but real, real slow. I come to find out from you what’s wrong.”

He said it all matter-of-factly with no seeming concern or emotion. His eyes were half closed and focused on the door behind me.

I looked at his wife, who was sitting on a chair in silence. She was a plain-looking woman in a simple housedress. Everything about her was colorless. She was not wearing any makeup; her hair was pale brown, streaked with gray; and her eyes were faded gray and looked to be without lashes. She just sat, rigid and unsmiling, hands folded in her lap, and watched me. She never said a word, either in response to my initial greeting, or on her own when I included her in the conversation, even though she had plenty of opportunity. I turned back to the patient. “Anything else bothering you?” I asked Fred.

“No,” he answered, “except I got a little trouble swallowing.”

“Anyone in your family with anything like this?” I asked.

“No,” he answered again, “I have two boys in their teens and they’re okay.” As we talked, I watched the

small brief twitches in his arms and legs and in his chin. There was no pattern to these fasciculations—just random quick bursts of activity, like lightning bugs in the evening sky.

The conversation lagged as I focused my attention on the fasciculations. Fred looked down at his arm. “It’s funny about those things,” he said. “Every time I look, they’re there but I can’t feel them jump.”

I went on to examine him in more detail. Sensation was normal and reflexes were hard to get. He moved about without difficulty but his rounded, sagging abdomen showed he could eat his dinner jut fine but the food wasn’t building any muscles. Later that morning, I ran an electromyogram. The result suggested that his motor neurons were degenerating. Not a surprise.

There it was. Gradually developing weakness, muscle wasting, and fasciculations—all this was easily observable and then corroborated by the electrical tests of the muscles.

I explained the findings to him and to his wife, “The problem is similar to Lou Gehrig’s disease but much less severe. Your breasts are enlarged, and that raises the possibility of Kennedy’s disease.” I gave them some basic information about the disorder and suggested that we do a genetic test to confirm the diagnosis. Fred wearily nodded agreement.

When I saw him several weeks later the results were back and, as I had suspected, were consistent with the diagnosis of Kennedy’s disease. Fred seemed little interested in this news, but his wife spoke to me, for the first time it seemed. Her voice was flat. “Does that mean there is some treatment to help Fred?”

I knew I had been avoiding what was the main issue for the patient. Neither Fred nor his wife cared much what the disease was; they wanted to know what it meant for them, and quite rightly so. “No, there is no effective treatment,” I answered. “I wish there were. Having the diagnosis only means we have a better understanding of what is happening. Unfortunately, there is nothing we can do to stop or even to slow down motor neuron degeneration.”

“I suppose he could have passed this on to our boys. . .” Fred’s wife half said, half asked, her face expressionless while she looked down at her lap.

“No, there is no chance that Fred could have passed it on to your sons. If you had a daughter, she

could be a carrier and then she could pass it on to a son. But father to son, it never happens." I decided to avoid any detailed discussion of sex-linked inheritance. "The boys are safe. This illness ends with Fred."

I thought she might smile with relief but she just nodded her head, still looking down, not at me or at Fred. I turned to him and put my hand on his shoulder.

"I'd like to see you, Fred, every three to six months so I can help you deal with any problems that might arise, like having more trouble with your arms and legs or more problems with swallowing. We may need the physical therapist to show you how to make better use of your arms."

"Okay," he said, after a long pause. "Then, like you said, I guess I'll see you in a few months." He got up slowly. We shook hands as he left. His grasp was soft, with little pressure. When I let go his hand, his arm dropped to his side. His wife made no move to shake my hand, just nodded at me without a word or a smile.

I saw Fred every six months for many years after that. He always said he was doing fine when I asked how he was, to which his wife always silently shook her head. We never talked about anything except his weakness and he would always answer questions in a passive and seemingly disinterested way. He never made eye contact with me. I graded the strength of his muscles at each visit. He got weaker, almost imperceptibly slowly. Working became more difficult, and he finally applied for disability.

Fred's wife was there each time. I never saw the two sons. She was sober as ever and always quiet, except to say at every visit, "I think he's weaker than the last time you saw him."

I knew she had to be worried about the future, although she never voiced it. Assuming this to be true, my answer was always, "His strength seems about the same, but compared to two years ago he does seem a little weaker." I silently hoped that her unvoiced concern about his ultimate incapacitation was unfounded.

As the years went by, I noticed that Fred's speech was becoming more slurred as a result of the increas-

ing weakness of his tongue and lips. Swallowing too was becoming more difficult. He had no trouble breathing, but he no longer had a strong cough. I worried that he might have problems with aspiration down the road.

My concern turned out to be groundless. To my relief, Fred never lost his ability to walk or to perform his daily activities. He just seemed to keep winding down—slowly. Although his speech became difficult to understand, he always managed to swallow without choking on food or liquids.

Twenty years after I had first seen Fred, I ran into his wife in the hospital corridor. I was making rounds and she appeared, walking swiftly and purposefully—with her head up. Her energy was amazing and for a second I wasn't sure she was the same person I knew. I was surprised she was alone—I'd never seen her without Fred. I suddenly realized I'd never learned her name. She stopped when she saw me. "Hello, is Fred okay?" I asked.

"No," she answered, "he's been here the last four days. He died all of a sudden this morning. I'm on my way to make the final arrangements."

I was dumbfounded by the news and amazed that she was so calm and showed so little grief. "Nobody called me to see him," I said, incredulously. "I had no idea he was in trouble."

"No, no, the problem wasn't his muscle disease," she said. "I thought, at first, it was. He was so short of breath—I was afraid he'd stop breathing altogether—and he had some chest pain so I brought him in as an emergency in the middle of the night. They said he had a heart attack and that it was huge. They did everything but he never had a chance." And, for the first time in twenty years, she looked directly at me and smiled—an unbelievably warm and friendly smile.

"You know," she continued, "all these years I lived in constant fear for Fred, worrying that he would end up an invalid, unable to care for himself, lying there unable to eat, unable to move, and having trouble breathing. I was sure his dying would be slow and awful. I didn't know how I'd manage. I spent the past twenty years in dread. And then this happened. It came out of nowhere. We never saw it coming."

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