Testimony Before the Public Health Committee
State of Connecticut General Assembly
Task Force to Study Rare Diseases
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Members of the Task Force, thank you for inviting me here, today.

Four years ago, my husband, Peter, was diagnosed with a rare disease—sporadic inclusion body myositis (sIBM). Despite my career teaching human physiology, I had never heard of it. I learned a lot real fast, and as I watch it manifest in him, I feel as if we're falling into an abyss. It is the most common muscle-wasting disease in those over the age of 50 and is more common in men than in women. It is considered an inflammatory disease of unknown etiology. There probably are about 100 sIBM patients in Connecticut. sIBM affects the extremities, and in some patients the cricopharyngeal muscle for swallowing, the diaphragm, the heart or the lungs.

Let me introduce Peter. Debilitating diseases are difficult for everyone, but they are particularly hard on those who've been active all their lives. He is a world-famous expert on herpetology and the identification of living reptiles and their skins and products. He ran the Reptile House at the Bronx Zoo for 30 years and finished his career as curator of the Central Park Zoo. He has an honorary appointment at Yale. His specialty is crocodilians and he has travelled around the world studying them. He still serves as a forensic examiner, mostly working for auction houses, such as Sotheby's and Christie's. You can imagine that not being able to ambulate well is hell for him, and our visits to Sotheby's and Christie's take a lot of pre-planning so he can minimize walking.

We knew there was something wrong. About 10 years ago, Peter complained of difficulty swallowing (dysphagia). His nephrologist, who had prescribed anti-hypertensives and a statin, waved our concerns away. But I argued with him, so he referred Peter for a barium swallow test that showed nothing. Our PCP ordered a blood test for creatine phosphokinase (CPK), a possible sign of muscle deterioration. The titer was significantly above normal and he immediately took him off the statin, which is known to affect muscle. We then saw a rheumatologist who suggested one of the muscle wasting diseases, but an antibody test was negative. There was no antibody test for sIBM then.

Next, Peter started to fall. He fell in the street, he fell in stores, he fell at home. When he realized that he could no longer grasp a hammer, we saw the rheumatologist, who ordered an MRI, informed him he had several herniated disks and suggested back surgery. No, I thought. There is a pattern here. I insisted he see a neurologist, who

administered an EMG and referred him to the Hospital for Special Care. It took nearly seven years to diagnose the disease. As I watched the surgeon remove a 1-inch-by-3-inch biopsy sample from Peter's thigh muscle, it hit me that this was going to be a long, hard slog and was not going to end well.

Although the disease itself may not be fatal, there is no treatment. Leg and arm exercises can slow its progression, but it can't be stopped. It is not uncommon for those patients with dysphagia to succumb to aspiration pneumonia, a constant fear for us. Even a slight fall can become a catastrophic injury if Peter fractures a hip or a long bone in his arms or legs. There's no rehabilitation possible because of the damaged leg and arm muscles. This terrifies us.

For months after his diagnosis, he had PT and OT twice a week, and several sessions with the respiratory therapist who prescribed swallowing exercises. He is supposed to do the exercises daily, but as he loses muscle mass, they've become too tiring. He couldn't handle the swallowing exercises at all.

Peter is now losing the ability to walk, to carry any weight with his arms or to grasp with his fingers. He can no longer swallow most green vegetables, even if well cooked. He can't swallow spaghetti, pudding or mashed potatoes without choking. He makes a horrible choking noise as he eats and he cannot eat anything without a beverage. He needs assistive devices for many ADLs, even buttoning and unbuttoning a shirt--and we would not be able to afford the large items if it were not for the VA. They gave him an electric scooter and installed a scooter lift in our vehicle. They installed two chair lifts, one to the first floor from the garage and another to the second floor. They paid for leg braces and they supplied him with a spiffy rollator.

One of the major issues I struggle with is that doctors usually are unfamiliar with sIBM. Most physicians my husband sees, other than neurologists and his VA PCP, are unacquainted with the disease. We try to educate them. But I have become a helicopter wife—the bad partner—peppering medical professionals with questions and feeling out their knowledge of the disease. One PT sent by the visiting nurses told us he had treated five sIBM patients in our small town—statistically unlikely for a disease that affects about 3.5 per 100,000 people. We cancelled PT immediately.

The year after Peter was diagnosed, he had a severe bout of vomiting. We were out running errands, so we brought him to the local hospital ER. He had cholecystitis. Hospital staff decided to remove his gallbladder without attempting to communicate with me or even tell me the doctors' names. It is imperative that any anesthesiologist involved in surgery on Peter speak with his neuromuscular physician because sIBM patients do not respond to muscle relaxants used in anesthesia. An anesthesiologist, seeing no response to the drug, will increase the dose, potentially threatening the patient's life. It was safest to treat him at major hospital that could better handle a rare disease patient. If it wasn't for the assistance of a friend, who knows the hospital's CEO, Peter would have gone into surgery. I demanded he be moved to Hartford Hospital, where the physicians understood the need to communicate with the neuromuscular

physician and Peter received safe and appropriate anesthesia. I am vigilant when Peter sees a new doctor and I am particularly vigilant with anesthesiologists or anesthetists, whose experience likely does not include sIBM patients.

Because I have taken over tasks Peter previously did, my life has changed radically. After it became clear that my teaching days were over, I realized that even if offered a class, I probably would turn it down. I worry about leaving him home alone because he falls. Peter drove me everywhere, so for 30 years, I hardly drove at all. Now, I drive because between his disease and his heavy leg braces, his leg gets too tired. I also do most of the household chores—tasks I hadn't done in years.

Going out to eat, shop or for entertainment depends upon the venue's accessibility. If it isn't ADA compliant, he can't go, and as he can no longer visit friends whose homes have steps leading into them, his ability to socialize is limited.

If he falls in public or can't get out of a seat in a restaurant or theater, we're completely dependent upon the kindness of strangers. I can't lift him, and even if I could, I'd need to be trained to do it. When I'm out with him, I feel helpless because I don't know when he'll collapse to the ground. Thus, I, too, am a captive of his disease. He doesn't need an aide yet, but he needs a babysitter so I can go out and feel secure. I'm not sure I can afford it, and when a friend would stay with him when I went to teach, he complained that she disturbed his work on a scientific paper.

I have two specific requests:

1) Medical personnel who are not specialists in rare diseases should be required to receive CME training about those statistically significant enough that they might see a patient who has one.
2) If federal ADA laws are nullified, our state lawmakers should make sure those laws are duplicated and modernized in Connecticut state law. Forcing disabled persons to stay within the confines of their homes and unable to participate in society is unfair and discriminatory.

Thank you very much for your time.