

Lori's Choice

IF YOU KNEW YOU were going deaf, what price would you pay for a few extra weeks of listening to Mozart, the sound of your lover's voice, or the wind rustling through the trees?

About a year ago, Long Islander Lori Davila faced such a terrible calculus. A benign tumor was pressing against her brainstem. The ever-enlarging growth had to be removed, even though it would mean certain deafness. But every day she waited increased the risk of neurological complications, or worse.

Lori, now 30, opted to have the surgery as soon as possible, rather than take the gamble. "But when I learned I would lose my hearing," she says, "I cried for about two months straight."

The operation also left her face partially paralyzed. Now, she faces multiple surgeries for new tumors in her brain and on her spine.

Such is the lot of patients with neurofibromatosis type 2 (NF2), a rare and incurable genetic disorder in which one tumor after another infiltrates the central nervous system. Typically, the auditory nerves are the first to be affected, starting in early adulthood. (In a more common sister disease, NF1, the growths primarily affect the peripheral nerves.)

Over the years, Lori has grappled with depression, yet she remains optimistic and good-humored, especially now that she's found a multidisciplinary team of specialists at NYU who are well acquainted with this "orphan" disease and can offer hope in the form of experimental therapies.

Like many other NF2 patients, Lori spent years searching for physicians who could provide any help. Most of the neurologists she visited hadn't even heard of the disease. "Anytime I saw a new doctor, I'd print out information on NF2 from Wikipedia for them to read before my visit," she says.

Lori's battle with NF2 started in college, when her vision began to fade. She was referred to a local neurologist, who discovered a tumor engulfing one of her auditory nerves. The tumor was removed, leaving her deaf in the affected ear, a common side effect of the surgery. Her recovery



Lori Davila and J. Thomas Roland Jr., M.D.

was complicated by a cerebral spinal fluid leak, necessitating a spinal tap and five extra days in the hospital. She struggled to complete the semester; dropping out meant losing her health insurance.

Lori had no idea what to expect in the months and years ahead. "The doctors knew nothing about NF, other than I had it," she says. "I didn't know where to go for information. So I just tried to be normal and got regular MRIs to make sure nothing grew too big." But the disease progressed, eventually affecting her sense of balance, disrupting her sleep, and sapping her energy.

About two years ago, her face started tingling and she began losing hearing in her good ear. Through a friend, she learned about the NF2 team at NYU. The timing

was fortunate. "Planning is critical for these patients," says J. Thomas Roland Jr., M.D., associate professor of otolaryngology and neurosurgery, who manages her care. "I've seen more than a few cases where surgery was delayed and removing the tumors caused substantial nerve damage, so much that remedies like ABIs or nerve grafts were no longer possible."

At NYU the tumor on her brainstem was discovered and removed. She has since undergone several other procedures at the Medical Center, including a nerve graft, reducing her facial paralysis, and an auditory brainstem implant (ABI), restoring partial hearing in one ear.

By default, Dr. Roland has become Lori's primary care provider. "No one else was

overseeing her care,” he says. “That’s when I had this idea that we should have a center where patients like her can get comprehensive care, the latest information, and proper counseling, as well as access to clinical trials.”

With a grant from the Children’s Tumor Foundation, NYU is establishing a center for patients with NF1, NF2, and a related disease called schwannomatosis, bringing together specialists in neurosurgery, otolaryngology, ophthalmology, pain management, audiology, rehabilitation, and counseling. The center, the first of its kind in the metropolitan area, will also coordinate basic and clinical research into causes and treatment of NF.

“I am still struggling to get back to a normal life,” says Lori, who stays in constant touch with friends and family and other NF2 patients via e-mail and instant messaging (which is how she was interviewed for this article). “It was very difficult to learn how to communicate. I lost my hearing months after getting married—as if married couples don’t have a hard enough time communicating! I pushed everyone—my family, my closest friends, my husband—to learn to ASL (American Sign Language). I refuse to be that quiet person in the corner left out of all conversations. With lip reading and my ABI, I can understand what people say, and it’s making it easier.”

Easier, but not easy. “I can get away as passably normal to strangers, but once I start to talk or smile, it’s impossible to miss,” she explains. “A lot of people treat you differently when something looks physically wrong with you.”

Although NF2 patients are living longer and longer these days, thanks to early and aggressive surgical intervention, the only effective treatment, Lori’s future is uncertain. “I have been deeply worried because of the number of tumors I still have,” she says. “I am hoping desperately I can get on some type of medication to prevent me from having to have more surgery.”

Several promising anti-tumor drugs are now in the research pipeline, and NYU is preparing to launch several clinical trials.

In the meantime, Lori soldiers on, “enjoying the little things that I can do and do have.” She looks forward to finding a job that can accommodate her disabilities and has started a small embroidery business to raise NF2 awareness and money for research.

“All these patients impress me overwhelmingly with their courage, fighting through this disease and grabbing the most out of life that they can,” says Dr. Roland. ●

The Music of the Night

To cure sleep apnea, an ancient instrument may be best medicine of all.

is indigenous to Australia, where it’s been used in traditional Aboriginal ceremonies for thousands of years.

Now, modern medicine has found a new use for this ancient artifact. With practice, the didgeridoo produces an eerie, reverberating bellow. But to those afflicted with sleep apnea—a potentially serious sleep disorder in which breathing repeatedly stops and starts—the sound is music to their ears.

“In people with sleep apnea, the airway intermittently collapses during sleep,” explains Dennis Hwang, M.D., a researcher in the Division of Pulmonary and Sleep Medicine. “We believe that learning to play the instrument strengthens the muscles of the upper airway and reduces the airway collapsibility during sleep.”

Yildiz, an information technology expert at Merrill Lynch, is part of a 10-person study being conducted at NYU to determine whether playing the didgeridoo regularly can help to cure their disorder. Sleep apnea (Greek for “without breath”) affects as many as one in five middle-age adults, who literally stop breathing for moments while they are asleep. These stoppages cause the brain to wake up, which allows breathing to resume, but the pattern may leave him sleepy and irritable during the day. Loud snoring is a common symptom of sleep apnea, although not everyone who snores has the disorder.

“Lately, I started feeling very drowsy and tired during the day and waking up many, many times during the night,” says Yildiz. “I couldn’t stay awake, and I thought, something is wrong.”

Recent studies suggest that sleep apnea increases the risk of high blood pressure, diabetes, heart attack, and stroke. Traditional therapy uses continuous positive airway pressure (CPAP), delivered through a cumbersome mask and tubing, to keep the airways open. But CPAP is not for everyone.

Playing the didgeridoo is likely to be a much more pleasant option.

In 2005, researchers in Switzerland reported in the *British Medical Journal* that playing the didgeridoo decreased daytime sleepiness, and the severity of sleep apnea, in people with the condition. In the NYU study, researchers will measure the patients’ airway collapsibility before and after lessons to document how much the instrument strengthens the muscles. The key, says Dr. Hwang, is probably the peculiar “circular breathing” technique, which allows the player to sustain a note almost indefinitely without pausing to inhale.

Instructor Giten Tonkov of the Energy of Breath Institute has been playing the didgeridoo for eight years. He still remembers the first time he heard its distinctive sound. “It was mysterious and mystical,” he says, “and the power of the vibration it gives out is very different. It struck a chord with me.” ●

