Dystonia

Heartbreak & Hope

A family touched twice by a little-known disease seeks answers and a cure

By Page Leggett
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In 2005, a game of backyard basketball altered the lives of seven-year-old Tyler Staab and his dad, Rick.

The two were playing when Rick noticed Tyler's errant right elbow was resulting in a lot of missed baskets. Rick demonstrated how to tuck his elbow in. When Tyler tried the same thing, his arm shook.

Rick recalled seeing Tyler writing with difficulty and wondered if the two issues were related. "We found out Tyler had a problem writing, but hid [it]…because he didn't want us to worry. He had taught himself to write left-handed so well that his teachers didn't notice either."

Something was wrong with Tyler. The Staabs set out to find out what.

The most common disease you've never heard of

While Rick and his wife, Michelle, had never heard of dystonia—the disease Tyler was ultimately diagnosed with—it is actually the third-largest movement disorder in the world.

Dystonia is a neurological movement disorder that causes muscles to contract and spasm involuntarily. In dystonia sufferers, muscles are working overtime. They don't relax as they should. Opposing muscles may contract at the same time, as if vying for control of a particular body part. This internal tug-of-war results in twisting, repetitive movements and awkward postures. No part of the body is immune. Limbs, torso, neck, face, eyelids—even vocal cords—can be affected.

While there are many different types of dystonia, the disease can be broadly divided into two main categories: primary (the type Tyler has) and secondary dystonia.

Mahlon R. DeLong, MD, scientific director at the Dystonia Medical Research Foundation (DMRF) in Chicago, explains the difference. "Dystonia is classified as primary or secondary based on what is known about the cause," he says. "A person with

Dystonia, the third largest movement disorder in the world, causes muscles to contract and spasm involuntarily. It is estimated that nearly 300,000 people in the United States and Canada suffer from dystonia and one-third of them are children.

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primary dystonia has no other neurological symptoms or disease, except perhaps for a hand or head tremor. Primary dystonia is genetic or presumed to be genetic and cannot be attributed to any other cause.

"On the other hand, secondary dystonia is attributed to a known factor or disorder," he continues. "Physical trauma, especially to the head; drug or toxin exposure; and other diseases and conditions—including Parkinson's, cerebral palsy and multiple sclerosis—can include dystonia."

Warning signs and causes

Although there are exceptions, dystonia generally develops gradually. Symptoms are often subtle and, therefore, easy to ignore. Warning signs can include small facial spasms, difficulty chewing, changes in speech, stiff neck and hand cramps while writing.

Dystonia can arise from a birth injury or can follow a brain infection such as encephalitis. While most primary forms of dystonia are thought to be caused by a combination of genes, research continues to identify other unknown factors. It is estimated that nearly 300,000 people in the United States and Canada suffer from dystonia, and one-third of them are children. The actual number may be much larger, as Dr. DeLong explains: "Secondary dystonias that are caused by a number of diseases and traumas are more prevalent than the primary forms. When we look at the dozens of diseases that can cause dystonia, such as Parkinson's, Huntington's and multiple sclerosis, the numbers of dystonia cases may be propelled into the millions."

Unlike other common neurological disorders such as Alzheimer's and Parkinson's the disease is not degenerative. Further, dystonia looks different in children than in adults. Jan Teller, PhD, science director of the DMRF, explains, "Children who develop dystonia typically develop severe generalized forms that affect the arms, legs, trunk, neck and face. Adults who develop dystonia typically experience symptoms in a specific area of the body such as the


Kim, diagnosed in 2000
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Pain can be a component

Dystonia can cause pain that ranges from mild to severe.

Dr. DeLong of DMRF says, "The pain level associated with dystonia can vary from none to excruciating. Some individuals liken the pain to an intense 'Charley horse' type muscle cramp that does not relent or go away." Tyler has severe and almost constant pain.

"In many cases, with treatment the dystonia symptoms, such as the tight, contracting muscles, will lessen, as will the abnormal postures, disfigurement and the pain," Dr. DeLong continues. "Pain may also be treated with oral drugs. Some individuals find relief through regular relaxation practices, and/or gentle massage from a professional who is familiar with dystonia."

Even the simplest actions can be laborious for people with dystonia. Tyler has difficulty eating because he cannot open and close his jaw normally. Since his muscles are constantly working, he burns a lot of calories even when he sleeps. A feeding tube in his stomach helps Tyler maintain a healthy weight.

After the diagnosis, now what?

Because there are so many different types of dystonia, there is no one-size-fits-all treatment. Treatment must be customized to each patient. Often, a combination of treatments is needed.

"Patients haven't consistently responded to one type of therapy, so multiple strategies are often recommended," says Dr. Okun. "These strategies should optimally include an interdisciplinary approach. In many cases, pharmacological agents, oral medications, botulinum toxin [Botox] and surgical approaches such as DBS [deep brain stimulation] may be used. Strategies must be tailored to fit the patient because every dystonia patient is different."

Dr. Teller of the DMRF says non-drug therapies may also be useful treatments. He describes these options as "adapting habits, physical or occupational therapy, braces and, in some cases, orthotics."

The surgical option

After much research, the Staabs learned that DBS, which has been successful for some Parkinson's patients, was also an option for dystonia patients.

In June 2006, Tyler bravely underwent DBS surgery. He was awake during the entire eight-hour operation. "Two electrodes were placed in his brain, and two control packs placed in his chest," Staab explains. "The doctor stimulated the area to see how Tyler's muscles would be affected. The electrodes can be turned on to stimulate a specific area of the brain. Brain signals can be disrupted in order for the patient to gain control over his or her movements. The whole system is then turned on, and the patient will need to be 'fine tuned' for the best effects."

Staab says that while DBS was initially successful, it did not take long before Tyler was worse. "In fact, it was only six months after the surgery. After many CT scans, it was determined that his electrodes had migrated with the growth of his skull. He had another surgery to add another lead, and it has helped. We can tell the DBS is working because Tyler's symptoms get worse if his system is turned off."

Medtronic's implantable neurostimulation device. Reprinted with the permission of Medtronic, Inc. ©2006

DBS is not for every dystonia patient, Dr. Okun cautions. "DBS should only be considered in a select group of patients with dystonia patterns known to respond to electrical stimulation. This therapy is only appropriate for a subset of well-screened patients. Research may allow expansion of indications within dystonia in the future."

A parent's nightmare

Knowing they needed to do something to help find a cure for their son and others suffering from dystonia, the Staab family founded Tyler's Hope for a Dystonia Cure, Inc., a local non-profit 501(c) (3) organization committed to funding pediatric neurology research to prevent and cure DYT1 Dystonia. Then, in 2008, while the Staabs were dealing with the day-to-day challenges of Tyler's dystonia, the unthinkable happened. Their second child, seven-year-old Samantha, began to exhibit symptoms of the disease. Unlike her brother, Samantha's legs were the first part of her body to be impacted.

"Although Tyler is my firstborn and the namesake of our foundation, my daughter is also fighting this horrible disorder with unbelievable grace and courage," Staab says.

There is a third Staab child. "Luke is four years old. We do not know if he has the disorder yet," Staab says. The Staabs decided not to test their children for the gene until they showed symptoms. "Even if you have the gene," Staab says, "it is possible that you may not get the symptoms."

Someday…a cure

"Tyler and Samantha's disease has a known gene and a known protein to target for a cure," says Staab. "If we can stop the symptoms, these kids can go back to living a normal life. This disease is not neurodegenerative."

Dr. Okun also believes there is good reason to hope. "For at least one form of genetic dystonia"—DYT-1—"there is tremendous enthusiasm that we will see either more meaningful treatments or a cure in our lifetime," he says.

Staab is hopeful—and determined. His foundation is playing a role in finding that cure. "Tyler's Hope has funded the writing of a book on the multidisciplinary approach to dystonia care that is being used at the Tyler's Hope Center for Comprehensive Dystonia Care. The book will be published this summer. This approach incorporates everything from neuropsychology to physical therapy.

"We are also funding drug discoveries that could stop the symptoms and return dystonia patients to normal functioning lives," Staab says.

As the Staab family had their lives altered by dystonia, they hope to radically alter—for the better—the lives of other families dealing with the disease. Staab adds, "We will find a cure; we just need to hurry."

UPCOMING EVENTS FOR TYLER'S HOPE

- **Tyler's Hope Texas Holdem Tournament in Gainesville, FL**
  Saturday, March 21, 2009
- **Tyler's Hope Wine Tasting in Chapel Hill, NC**
  Thursday, April 16, 2009

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