The Miracle of Nibs

The following is an account from Alison Frase, Co-Founder of the Joshua Frase Foundation.

The Joshua Frase Foundation (JFF) raises funds for medical research for the treatment of and/or cure for neuromuscular disorders, to increase awareness of these diseases, and to build a network of support for families affected by these devastating disorders. JFF supports doctors at Boston Children's Hospital, Harvard Medical School, and the Wake Forest Institute for Regenerative Medicine. Each institution is making great strides towards finding therapies to treat and potentially cure neuromuscular disorders. When a therapy is proven, thousands of sick children will be able to receive these treatments, which could extend and improve the quality of their lives.

This may be the last chance to save my son Joshua. His health has declined dramatically and time is running out. It is a miracle that he is now 14, but it has been a tough year.

The Food and Drug Administration (FDA) requires that doctors show success in pre-clinical trials using a large experimental model, before considering therapies to be tested in humans. Testing on this type of model for MTM provides a critical opportunity to develop and test treatments for safety and efficacy before running human trials on children like Josh.

And so our search began…

In the fall of 2008, a female Labrador Retriever was discovered to carry the same gene as I do for my son's muscle disorder called mytubular myopathy (MTM). To date, this was the first MTM large animal ever discovered by researchers anywhere in the world. This dog, Trixie, was discovered by a veterinarian in Canada by the name of Dr. Elizabeth Snead. Trixie did not show any signs or symptoms of the disorder. The confirmation came when Trixie gave birth to a litter of puppies and two out of ten showed symptoms of MTM. The muscle tissue from the pups was sent to the University of California, San Diego, to Dr. Diane Shelton, who confirmed they were born with a muscle disorder.

Before we could speak to Trixie's owner, she was spayed and our promise for research with a large animal model came to a screeching halt! I shared with Dr. Snead where we were with research and how we desperately needed a large animal model for pre-clinical trials. Determined, Dr. Snead spent weeks locating a female sibling of Trixie named Nibs. Dr. Snead learned that Nibs also gave birth just months before and two out of her ten pups were affected, which led us to believe that Nibs was a carrier of the same gene.

Dr. Snead provided me with the number of Nibs' owners, Vic and Karen, who also live in Canada. I called Vic, not knowing to what extent, if any, he would consider getting involved. I explained where we were with research and what we needed to do to get to clinical trials. I told him that time might be running out for Joshua. I told him about the nine children who had died this year due to this neuromuscular disease. Before I could finish, Vic said, "I want to give you this dog, I want to help your son." I was overwhelmed with great joy. His kindness was beyond words.

My husband, Paul, and I agreed that I needed to get on a plane immediately. We felt the urgency to get the dog as quickly as possible. Vic and Karen adjusted their schedules and drove three hours one way to meet me at the airport in Saskatoon, Canada with Nibs. It was an emotional meeting and a very touching goodbye. Nibs and I bonded quickly and by morning, I felt she trusted me.

The trip itself felt symbolic of my journey with MTM. From the moment I stepped onto the first flight I experienced setback after setback. Ten ticket changes and four airlines later - I delivered Nibs to our researchers in North Carolina. Everything about this journey was a lesson in perseverance. My guard was up the entire time - just like it has to be with Joshua. On the second flight change, the airport lost my luggage and Nibs' carrier - so we were scrambling around in Saskatoon to not only beat the impending snowstorm but also find supplies for the trip back home. Although the trip was fettered with frustration on my part, and confusion on the airlines part, there were also moments of extreme joy and gratefulness. The instant bond with Nibs felt like she and I were created to know each other, and that somehow she just knew that she was on a very important mission. Through all the chaos at the airport, Nibs remained calm. Vic and Karen have been like so many others I've met on this journey - willing to sacrifice what brings them joy so that my son and others like him have a chance at life. Leaving Nibs at the research center felt like I was leaving a piece of my heart there too - because I was. It was emotional, to say the least. I drove away with a hope I've seldom dared to have on this journey with MTM - praying that my son would live long enough to experience the benefits of those crazy three days.

I sensed a new chapter in our journey, another step towards finding a cure for Joshua and many other children like him. We've entered another stage. We've crossed another barrier towards FDA approval. Hope is finally in sight.

Shortly after arriving in North Carolina, Nibs became pregnant and delivered 12 puppies. Six of the puppies carry the gene for myotubular myopathy. The pups are doing extremely well, are happy, eating up a storm and getting really big. The staff at Wake Forest Institute for Regenerative Medicine is absolutely amazing, giving the pups extra care and attention seven days a week.

Once Nibs' duties as a mother were fulfilled, and after thoughtful consideration, Paul and I decided it would be best for Nibs to return home to her family. In Canada, she resumed her life on the farm where she runs daily with the horses and hunts wild rabbits. As I walked through the airport in Saskatoon later that summer to deliver Nibs, I witnessed Nibs' joyful reaction when she saw her owners, Vic and Karen. All Vic had to do was whistle, and Nibs leapt towards him, confirming that Paul and I had made the right decision.

As I write this, I am on my way home from returning Nibs to her family, confident that we are one step closer to finding a cure for neuromuscular disorders.

The past several months have been surreal – for the first time ever a treatment for myotubular myopathy may be within reach. I am so grateful to Nibs for the 12 gifts she has given us. Wake Forest researchers are learning more about MTM and neuromuscular diseases from these dogs that carry the gene than they ever have before. Knowledge gained from these animals may one day lead to treatments not only for MTM, but other neuromuscular diseases. It will be a miracle for our son Josh and thousands of children like him if our goals are achieved.

Thank you for your interest, your hope and your willingness to participate in finding and funding this miracle.