



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 and/or cure for neuromuscular disorders. Our goals are to increase awareness of these diseases, and to build a network of global support for families affected by these devastating disorders. JFF supports doctors at Boston Children's Hospital, Harvard Medical School, Wake Forest Institute for Regenerative Medicine and the University of Washington. Each institution is making great strides towards finding therapies to treat and potentially cure neuromuscular disorders. When a therapy is proven, thousands of critically ill children will benefit from these treatments, which could mean extending and improving the quality of their lives.

I wrote this story as Joshua was in a decline and I felt it was important to take you to this moment in time.

This may be the last chance to save my son Joshua. His health has declined dramatically and I am sensing impending danger. It is a miracle that Joshua is now 14, given the odds he would never celebrate his first birthday. It has been a tough and critical year physically for Joshua.

We knew the gene replacement therapy success we were showing in the mouse model had to translate to a larger animal before we could even consider getting the FDA's attention for human trials.

And so our search began...

In the fall of 2008, a female Labrador Retriever was discovered that possibly carried the same gene as I do for my son's muscle disorder called myotubular myopathy (MTM). To date, this was the first potential 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 the tissue sample was presenting like MTM, but it was still speculation without a confirmed genetic diagnosis. At that point, I pulled back to let the two institutions pursue engaging with the owner about their potential involvement.

Six weeks later, before the research team could speak to Trixie's owner, she was spayed and our promise for research with a large animal model came to a screeching halt! It was early December 2008, 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 nearly 2 weeks on the hunt across Canada locating owners of distant relatives of Trixie who were also having affected litters. Dr. Snead received a call back from a cowboy in the middle of Canada who had a female lab named Nibs. Dr. Snead learned that Nibs also gave birth just months prior and two out of her ten pups were affected with an unknown disorder, which led us to believe that Nibs could possibly be a carrier of the same gene.





Dr. Snead provided me with the number of Nibs' owners, Vic and Karen. 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 son Joshua. I told him about the nine children our community had lost this year due to this tragic neuromuscular disease. Before I could finish, Vic replied, "I want to give you this dog, I want to help your son." I was overwhelmed with great joy. His kindness was beyond words and for the first time in thirteen years I had the promise of hope.

My husband Paul reminded me it was uncharacteristic for me to give up the reigns of retrieving the first dog. He then told me I was to get on a plane to retrieve the next MTM dog and I totally agreed. He reminded me of my "never take no for an answer" mentality, a quality he often credits to my New York upbringing.

Vic and Karen adjusted their schedules and drove three hours one way to meet me at the airport in Saskatoon, Canada with Nibs on December 27, 2008. It was an arduous journey to say the least. I arrived in customs without my luggage or dog crate that was nowhere to be found, on the eve of an impending crippling blizzard. It was an emotional meeting and a very touching goodbye for Vic and Karen. Vic and Karen have been like so many others I've met on this journey – individuals willing to sacrifice what brings them joy so that my son and others like him might have a chance at life. Nibs and I bonded quickly and by morning, I felt she trusted me. The next morning I awoke scrambling around in Saskatoon to not only beat the impending snowstorm but also to find supplies for Nibs for the trip back home.

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 researcher 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. 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 for me. Through all the chaos at the airport, Nibs remained calm. Leaving Nibs in the hands of my researcher 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. Five of the eight female puppies carry the gene for MTM and one of the four males was affected. The pups are doing extremely well, are happy, eating up a storm and getting really fat. 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, I decided it would be best for Nibs to return home to her family. As I walked through customs in Saskatoon airport later that summer to deliver Nibs, I witnessed Nibs' joyful reaction when she heard Vic whistle from across the crowd. Nibs leapt towards Vic and Karen through the crowd, and in that moment I knew I had made the right decision. She will resume her life on the farm where she will run daily with the 25 horses and chasing wild rabbits.

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 this tragic and fatal neuromuscular disorder.

The past several months have been surreal – for the first time ever a treatment for MTM 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 this tragic neuromuscular diseases from these invaluable animals 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 Joshua and the Joshua's of the world if our goals are achieved.

I hold tight to the vision of hope planted in my heart early on in this journey; that there is a beautiful story to be written... ..but I had no idea of how this next chapter would unfold.