

Journey to a Cure

*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 the disease and to build a network of support for families affected by these devastating disorders. Great strides are being made towards finding therapies to treat and eventually cure neuromuscular disorders. When success 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 15, but it has been a tough year.

The Federal Drug Administration (FDA) requires that doctors show success in clinical trials from a large experimental model. 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 that carries the same gene as my son's muscle disorder, myotubular myopathy (MTM). To date, this was the first large animal ever discovered by researchers anywhere in the world, that expressed this disorder. This dog, Trixie, was found by a veterinarian in Canada, 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 that presented like MTM on a slide. 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 to get to clinical trials. Determined, Dr. Snead spent weeks trying to locate a female sibling of Trixie. She finally found her and her name was Nibs. Dr. Snead learned that Nibs also gave birth in August of 2008, and two out of her ten pups were affected, which confirmed to our researchers that Nibs was probably a carrier of the same gene.

Dr. Snead provided me with the phone 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 son Joshua. I shared with him about the nine children who had died earlier that 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 compassion 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. On December 27th, 2009, 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 a heartrending meeting; this family was willing to give us their beautiful family dog of four years to further our cause.

Nibs and I bonded quickly and by morning, I felt she trusted me. The trip was just a nightmare. The airlines lost my luggage along with the dog carrier. The airlines warned me of an expected snow storm later that day and of the drop in temperature; so I had to respond quickly. The next morning, Vic and Karen drove Nibs and me to a local pet store to purchase a dog crate. In the airport lobby, Vic frantically assembled the dog carrier as we were trying to make it through customs, and make the flight before I was stranded in Canada for a week. My guard was constantly up; looking out for Nibs' safety and her care. We stopped in Minnesota to break up the trip because I had no clothing for myself, but more importantly, no food for Nibs. This layover kept Nibs from sitting hours in a crate between flights.

The following day, Nibs and I arrived at the airport for the remainder of our trip. The airlines nearly separated us on two different flights, but after throwing quite a scene, they pulled Nibs off the one flight putting us together, and we arrived safely in North Carolina. Upon arrival, I handed Nibs over to one of our researchers. I was very emotional. I was torn between knowing what Nibs needed to do for our son, but I also knew that she belonged with me. There was this unexplained connection between Nibs and I, as I believe she knew what she needed to do for me. On my way back home to Florida, I called and told the researcher that after Nibs gave them a litter, I would like for her to come live with our family. I was assured of Nibs' safety and comfort throughout this process and we decided to place Nibs at a breeder's farm where she was surrounded by 18 other Labrador Retrievers, playfully enjoying her daily life.

My researcher spoke eagerly of the progress he believed we could make with Nibs' help. I sensed a new chapter in our journey, another step towards finding a cure for our son Joshua and the Joshua's of the world. We've entered another stage and 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 on April 15, 2009. 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. We wanted Nibs to resume her life on the farm where she could run with the horses and hunt wild rabbits daily. Nibs' excitement upon coming through customs when she saw her owners, Vic and Karen, confirmed that we 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 puppies she has given us. Our researchers are learning more about this neuromuscular disease from our dogs than they ever have before. I am confident that the knowledge gained from these animals will one day lead to treatments not only for MTM, but other neuromuscular diseases.

