

# GRACE'S STORY

## *A Little Girl's Battle With Autism & Mitochondrial Disease*

By Shannon Kenitz



Like all expecting parents, we were hoping to have a healthy child, and with that, the joy and dreams we once had for ourselves, and the dreams our children would find for themselves.

After the birth of my first daughter, Lily, I felt that nothing could ever come close to the feelings I had for her. She was my first child, and we went through so many “firsts” together as Mother and child. Like most moms, I started feeling the “Maternal Itch” to give Lily a brother or sister with which to play. When Lily was 2, we found out we were expecting a baby girl, whom we named Grace—a name that has come to mean more to us than we could have ever imagined at the time.

Grace was born a healthy child—or so we thought. We left the hospital in the usual amount of days for a normal delivery, and headed home to raise our family, having no other cares in the world than feedings and changing diapers.

And so our story began. After bringing Grace home, it wasn't long before I thought that something seemed wrong with her. Grace wasn't tracking objects like Lily had at that point, and Grace had a lot of irregular eye movements, a rapid pushing of her eyes in a back and forth movement. I also noticed that she seemed to have difficulty with her feedings and wasn't able to latch on while I was breast feeding.

I made my first call to our family doctor and he thought that I wasn't trying hard enough with the breast feeding. I wasn't a young mom, by any means, having had Lily at age 30 and now Grace at 32. With Lily, breast feeding continued well into the age of two, and I knew what to expect. Thus, with Grace, I knew that there



was “something more” than improper latching, lack of persistence, or inexperience errors.

The doctors asked us to come in for some blood work, and the test found that Grace had very high bilirubin. In layman's terms, Grace was severely jaundiced. After a month of treating her jaundice and several trips back and forth to the lab, Grace's bilirubin levels finally normalized. We went on our way, naturally thinking that our lives would get back to normal.

However, days passed and Grace still did not want to eat and still was not interested in tracking objects. I once again made a call to the doctor, only to get a patronizing, “Grace is okay, Mrs. Kenitz. You're over reacting.” It became a routine.



Two months had passed, and with over several dozen attempts to get someone to listen to me, I finally took things into my own hands. I made one last appointment to see our family doctor and took with me a journal I had started to keep, recording the dates and times of all of strange Grace's eye movements, her dietary intake, as well as the daily routine, listing what Grace did and did not do. With me, I also took a book that outlined the milestones through which children generally progress, mainly as a reference tool.

I remember thinking to myself that I had to find a way to get these physicians to listen to me. I constantly had the feeling that they thought me incompetent, as a mother and Grace's advocate. In their view, the best thing that could happen was proving me wrong. And, part of me wanted to be wrong. But, if I was right, then the worst thing that could happen would be proof of their neglect—dismissive care that had been damaging my daughter. Their condescension was no longer acceptable to me.

After listening to me plead my case for over an hour and reading my journal, the doctor finally agreed to refer Grace to a specialist, not because he felt that anything was wrong with Grace, but more so to appease a ranting mother—me.

We were referred to a Neurologist and an Ophthalmologist. I finally felt some relief—something was going to be investigated. Yet, with these new developments, came new

fears. I started to really get nervous, and the two weeks before Grace's appointment seemed like a lifetime.

At this time, Grace was now almost 3 months old, she had lost two pounds, still was not tracking or eating well, and her rapid eye movements seemed to have become more frequent. Lily was not enjoying her baby sister. As did most of the family, Lily wondered why Grace never cried and why her little sister never looked or smiled at her when she would try to play with her. Obviously, this was not what we had envisioned for our family during first few months at home. As the appointments drew near, so did our fear of the unknown.

Our first appointment was with the ophthalmologist. Little did we know back then that this doctor would play



such an important role in our lives, just for merely believing us and for literally 'seeing' what no other doctor wanted to see. The doctor examined Grace for over two hours. He read my journals and more importantly he listened. While we were in his office, Grace started to do her usual eye rolling followed by a blank stare—a stare that was cold and distant.

After witnessing Grace's eye spasms, the doctor said that what we were seeing in Grace looked like seizures. He felt that we needed to meet the Neurologist directly at the hospital, instead of at the clinic's scheduled appointment time. The doctor made a few calls and we were out the door, headed for the hospital.

When we arrived at the hospital they were waiting for us. They rushed us into a private room. We could feel the weight of the whole situation. Something was terribly wrong with Grace. The neurologist explained to us that Grace needed to have an EEG (Electroencephalography) and MRI (Magnetic Resonance Imagery) as soon as possible, and that we needed to stay calm. I remember thinking to myself, "CALM?—That is what I have been doing until now!" Now my fears were materializing before my eyes—something was really wrong with my baby.

After several hours of tests, Grace was admitted into the Intensive Care Unit, and it seemed like hours before we got any answers. The neurologist broke the news that Grace was suffering from seizures, and that they would need to give her medication to help control them. I had no idea what these things meant or what this would mean to Grace and to our family.

Looking back to that day, it all seemed so unreal—that at one moment we had all our dreams coming true, and, in the same instant, with the proclamation of a doctor we had only

known for about 15 minutes, our lives began to unravel.

At first, the doctors felt that Grace suffered from a seizure disorder known as Epilepsy. I was okay with that—I had a friend who had Epilepsy and she was alive living a normal life. After several attempts to get the seizures under control and after 3 different seizure medications failed, we were taken to the University Children's Hospital for a second opinion—something we did not know about until after Grace was transferred there. This time we were given a new diagnosis: "Infantile Spasms to the Brain." Even the title sounded gloomy and horrible. This meant that Grace's seizures had become worse and that they were being caused by an underlying neurological disease, a disease that was just as mysterious as the symptoms she exhibited.

Grace was immediately placed on the top of a waiting list for a drug involving ACTH (Adrenocorticotropic Hormones) injections. The injections were supposed to help control her seizures so as not to cause any further brain damage. I realized that Grace had been having seizures since birth, and because they had gone untreated for 3 months at that point, they now had caused further brain damage to my little girl.

Grace continued to fail at the hospital. At 5 months, a feeding tube had to be placed in order to help her gain weight. Grace remained in the hospital for the next year, and the stress on our family continued to build. Grace started to have many other internal problems with bone marrow and blood problems. Leaving Grace for even a second was never a choice.

I remember the neurologist telling us that there had to be an underlying disease for there to be 'infantile spasms to the brain.' I realized that I was now in the fight of our lives to push the doctors to find this disease so that Grace wouldn't die.

Grace was then one year old and weighing just a little over 8 pounds. Even with the feeding tube, she was still considered as a 'failure to thrive' baby. She had undergone more blood transfusions than I thought possible, and her tiny body was starting to shut down. More importantly to me as a mom, Grace was a child who was not present. She had no idea how much she was loved, and all I wanted to do was hold her and tell her everything was going to okay—but even I couldn't convince myself that all would be well.



Up to that point, Grace had lived most of her life in the hospital, being transferred from The Mayo Clinic to New York Children's Hospital. Our old life was now only a

faraway dream. My older daughter, Lily, and I spent our days in the hospital, never leaving Grace's side. Lily's devotion earned her a doctor's coat with "Lily" embroidered on it.

On Grace's first birthday, the hospital threw a party for Grace. At our Neurologist request, she asked that all our family and friends come so that she could explain to them what was happening to Grace and what we should expect to happen. What was supposed to be a day filled with cake, balloons and presents now turned into sadness and questions from family and friends. Was Grace dying? I remember just looking down as the tears rolled down my cheeks. NO! NO! This is NOT happening! It can't be happening! But, it was. As my family and friends listened to the doctor's words, the room went silent. The verdict was that Grace was indeed dying, and that we needed to prepare ourselves and that the family needed to be supportive of me. The doctor said that Grace wouldn't live to celebrate her 2nd birthday.



I left the room and had a feeling off desperateness in my body. I felt that I couldn't breathe. I regained my composure and went back into the room. I told everyone that I didn't want to hear that word again. I told them that Grace needed our love, not our sadness. (To date, it has never been mentioned to me again, but I know that my family and friends talked about it frequently.)

Grace continued to fight for her life. The doctors were still searching for answers, always fighting the symptoms, never knowing the underlying cause. Grace was then a year and half old and had spent very few days at home with us. She was blind, still failing to thrive, and was having blood transfusions weekly. She was unable to regulate her own temperature and was having various biopsies to try to find the cause of her failing health. Grace was unable sit up, crawl, or hold anything in her hand. Yet, Grace was a fighter and every time her heart rate started to crash, she came back to me. Grace always looked so peaceful. She was so beautiful despite her body failing.

By this time we had many misdiagnoses, and we were transferred to Mayo Clinic to have a brain biopsy performed by a world-leading pediatric surgeon. It was during this time that Grace's muscle biopsy finally resulted in a diagnosis. The Muscle biopsy from Buffalo Children's hospital proved that Grace did have a neurodegenerative disease, a disease known as Mitochondrial Cytochrome C Redutase. There are several known forms of mitochondrial diseases, and

unfortunately Grace's is one of the rarest. Furthermore, the genetic neurologist could only find four other cases ever cited in journals, and all these children had died before the age of two. We got this news less than a month before Grace's 2nd birthday.

Although we finally had a diagnosis, we were still lacking a prognosis to help Grace. Once again our family was put to the test. It had taken almost 2 years to find the disease, and for all practical purposes, it still made no sense to us. It only put our family at odds against each other—each of us deeply rooted in what we believed was the right course for Grace.

After leaving Mayo Clinic we set out to see 'experts' on this disease. With so many different drugs and therapies affecting Grace's body, she continued to shut down. Grace had some good days, but the bad days had a way of taking over. Things became so bleak that the doctors recommended that we sign a DNR (do not resuscitate) order for Grace. I realized that we had little time left.

It took me less than 12 hours to reverse the dreaded DNR request. I started to feel that all too familiar panicky feeling with the shortness of breath. I had to do something—something that could change this horrible course of events.

I started to research a therapy known as Hyperbaric Oxygen Therapy (HBOT). I had read about HBOT in a Mothers united for Moral Support Newsletter and even though there was no mention of mitochondrial cases being treated with this therapy, Grace did have many similar characteristics to the Cerebral Palsy children in the newsletter who were being helped with this hyperbaric medicine.



I spoke to Grace's team of doctors, and they all laughed. They said it was a therapy that was unproven and not to waste my money. Nonetheless, I kept the idea of trying hyperbarics in the back of my mind. I thought, "All the parents I had spoken to, whose children were receiving HBOT and who were seeing improvements, couldn't be lying."

I started to do the leg work to find a center willing to treat Grace. I knew that I had exhausted all the usual medical means. The hospitals were no longer willing to fight for Grace because her disease was deemed terminal. They felt that I needed to "let her go." I decided to raise money myself for the hyperbaric therapy.

Grace was 3 years old when I took her out of the hospital, against medical advice. Our family made the cross-country trip to reach the hyperbaric facility. It took 4 hospital stops along the way, and when we arrived Grace was once again admitted into the hospital. She needed to be stabilized



enough to start the hyperbaric treatment.

While in the hospital, Grace was given a SPECT (single photon emission computed tomography) scan. This scan of the brain shows the flow of oxygen throughout the brain. It revealed which parts of Grace's brain were not receiving enough oxygen. The neurologist who examined the scan said that Grace's brain was in "bad shape." He agreed with her other doctors and recommended that we take Grace home and spend what little time we had left with her.

Having made the trip halfway across the country and raised the money (at great hardship to family and friends), we were determined to see Grace through hyperbaric oxygenation. This was our last hope, and Grace's last chance for life. I wanted so badly to believe that the therapy was going to work, but I also knew that Grace had so much going against her.

To be honest about the whole thing, one of the reasons that I pushed so strongly for hyperbaric oxygen was that it was the only therapy I had researched and not yet tried. I needed to know that I had tried everything possible before I could 'let her go'. I had no clear indication either way that it could even help or change anything at all. All I knew was that we had to try.

Grace started with a 2-treatment per day protocol, 6 days a week. What happened in the first four weeks was mind-boggling. Grace seemed more alert and was starting to reach for objects. She had been blind up until then. After 40 treatments of HBOT, we decided to go back to the neurologist who initially performed the first SPECT scan, and asked him to do a follow up scan. The findings showed "significant improvements."

For the first time in 3 ½ years, we were finally getting some positive news. The HBOT was working! Areas of Grace's brain which had not been getting enough oxygen were now being oxygenated, and her brain was starting to repair itself.

Clearly the hyperbaric oxygen started a change that has had a profound effect on our lives. Grace before HBOT and Grace after treatment are two completely different children.

PRE –HBOT: At 3 years old when we began Hyperbaric Oxygen Therapy.

Grace was blind, suffering from seizures, a.k.a. infantile spasms to the brain. Grace was fed only through a feeding tube and was still miserably failing to thrive. Grace spent most of the first three years of life in hospitals across the country, having had many muscle biopsies, a rectal biopsy, an eye biopsy and a brain biopsy that almost took her life. Grace was kept under a heat lamp since she was unable to regulate her own temperature. She had more blood, albumin and platelet transfusions than any one person should ever be required to have. Grace was unable to sit up, crawl, walk or talk. Grace was a child who was not 'present'—she always looked through us instead of at us. Grace had no idea that I was her mother.

POST- HBOT: Grace at 8 years old—nothing short of a miracle.

Grace is no longer blind. She is no longer having seizures and is off all seizure medications. When Grace was 5, her feeding tube was removed (by me because no doctor would take it out); she now eats on her own and loves food. Grace is no longer failing to thrive and is in the normal range on her growth chart—a chart whose parameters she had never before even remotely appeared to reach. Grace's body is now able to regulate its own temperature, so no lamps are needed. Grace's blood problems have diminished, and she has not required a transfusion since we began hyperbaric therapy. Grace sits up, crawls, and talks. At seven years old, my daughter surprised us when she stepped out of her beautiful little pink wheel chair—she has been walking ever since. Today, Grace goes to school. When I walk in the room she looks at me instead of through me. She knows I am her mommy—something that I never thought would be possible.

Hyperbaric Oxygen Therapy has not only saved my daughter's life, it has saved our family's life. Life before this treatment was filled with cold hospital rooms, needles, machines, constant ups and downs, never knowing if any one day would be Grace's last. Our daughter never had the normal first 5 years of life that a child should have. Hyperbaric therapy gave her a chance at living a normal life for a 5 year old little girl. When Grace became no longer medically fragile, she has been able to attend school, have friends, and join in friends' parties, sleep overs and camping trips. Today, Grace continues to go on class outings and has play dates with classmates. Grace is learning how to ride a bike and learning to dance.

The most amazing part is that Lily, now has a sister with whom to fight (yes, even this is a blessing), a sister to whom she can read and with whom to share secrets. Lily and Grace have the most incredible bond, and when Grace laughs, you would hardly imagine all that she's had to overcome. My parents now have a granddaughter to spoil and guide.

As for me, I now have all the hopes and possibilities that I did for her on March 6, 1999—the day Grace was born. I can now dream again without the nightmares. I no longer ring a bell before entering her room to make sure she is still alive. I have plans, a lot of plans for Grace, Lily and me. I look forward to the mornings of each new day. I no longer live in a panic and I no longer fear the unknown.



Our lives have been changed forever. I now have a new career as the Executive Director of the International Hyperbarics Association, Inc. (IHA), which is a non-profit association that promotes HBOT through education and research. I travel around the country speaking at conferences about Grace’s life and about how HBOT has played a role in her recovery. I receive hundreds of calls and letters from parents who have heard Grace’s story and who want to thank me for giving them the strength to go on. Someday Grace will read those letters herself and someday she will be writing chapters in her own book. That will be a great day.

It is true that we really never know when our time is up. I have known for many years that Grace’s time was not yet come. Grace is alive because of the Hyperbaric Oxygen Therapy. Her life now has much more meaning in this world than just being my daughter. Grace’s story inspires families across the nation to not give up hope. Her hope has given hope to families—families like ours who are benefiting from hyperbaric medicine.

In the final analysis, I think Grace’s story is about life itself. No matter what comes our way, we need to face it with hope for our loved ones, even when everyone around us has given up hope. Whether it is hyperbaric therapy or another kind of therapy, we as parents need to keep a motto of ‘HOPE AND POSSIBILITIES’ alive for our kids, because without hope you don’t have possibilities. If I have learned one thing through all this, it is that HOPE is the key ingredient to all the future successes of our children.

Grace is continuing Hyperbaric Oxygen Therapy and will continue to do so as long as we continue to see benefits. Grace may need this therapy her whole life, but I look at it like this: If Oxygen is Grace’s drug of choice, then I am the happiest mom alive because nothing is more elemental than oxygen.

We still live each day as if it could be our last together, and when I kiss Grace and Lily good night, they know how much they are loved. Ironically, the best part of my life so far is having a child with special needs. It has taught me to be kinder, more patient and tolerable of others. I would not

trade having Grace for anything in this world. Hundreds of other moms who have special-needs children know what I am talking about when I say that my life is truly fulfilled.

Having a special needs child gives us an opportunity to expand who we are in life—it gives us the creativity to learn how to juggle a thousand things for the sake of someone else. Instead of having one career, we have a multitude, that is, without the “validating” stamp of college degrees. I am a physician some days, a nurse others, and I have been a respiratory therapist, physical therapist, occupational therapist, and even a speech therapist on Tuesdays and Thursdays. I am a daily nutritionist, chemist, and researcher, and with all this spare time, an on-call pharmacist. As a public servant, I am an educator of parents (and sometimes doctors), a grant writer, and have even been a politician-of-sorts. Of course, just for fun, I find myself to be an adequate insurance expert and financial advisor—often having to find money from out of nowhere to pay for medical bills. In the end, my favorite title is that of “Mom” to two very special girls. I wouldn’t trade one minute of our lives together. At the end of the day, I have two special needs children because they are both “Special” in their own right and we all “need” one thing: One another. When all is said and done, nothing can take that away.

Keep HOPE alive. With hope comes possibilities, and when we have hope, anything is possible.

Shannon, mom to Lily & Gracie