

# Olivia

THIS IS OUR STORY—SO FAR.....  
THIS IS A PERSONAL STORY SHARED BY A FAMILY WHOSE CHILD  
HAS BETA KETOTHIOLASE DEFICIENCY (BKT)

It all began on February 1st 2005 when Olivia Belle Quinlan (who we call OOMPA) was born at approximately 9:00 PM. She was a perfect 8lbs 15 oz beautiful baby girl. Such a wonderful delivery and incredibly happy time for my husband Dan and I.

Two days after we arrived home from the hospital with our 3rd daughter Olivia Belle Quinlan I received a call that would change our life forever. It was around lunch time on Saturday and my older girls Cassie 5 and Chloe 3 were out with Papa. My husband Dan was working. I was sitting on the floor changing Olivia's diaper when the phone rang. I couldn't get to the phone but could hear a message from a local Pediatrician to call his office ASAP regarding the New Born Screening test results from the State Lab for my daughter Olivia.



When I spoke with the Doctor whom I never before met, he told me I needed to come to his office quickly and bring Olivia. He would not specify exactly why, but that her New Born Screening tests were positive for a disorder and we would need to be seen promptly. I never really understood or thought twice about the routine tests performed on newborns while in the hospital. I immediately panicked when he would not give me any information and feared the worst! I called my husband Dan who raced home to take us to the doctors. My mother had also been called and immediately left work to come with us. When we arrived at the Doctors office, he would not see us right away and when he did finally speak with us he was again vague and unspecific as to what she had, he said "he has only seen such a thing twice in 22 years" it's rare and can be life threatening. He had a terrible ego and bedside manner and was not comforting to us at all! He was by far the rudest person we had ever met, and unfortunately the one to deliver such heart breaking news. By this point I had already been crying since we left the house as I feared the worst. My whole world was turned upside down. I pleaded with the doctor to just tell me what the name of the disease was so Dan and I could begin searching for answers. He finally wrote down the name and we were on our way to the local hospital for a repeat test to confirm results were accurate. All we knew at this point was that she potentially had something very serious, but still didn't know what that meant to her little body. As we went to the hospital I sobbed like I have never cried before. It was like my world was closing in and there was nothing I could do. I could not understand how a perfect little girl who looked and acted so well could have something so seriously wrong with her. When we arrived at the hospital we did the blood tests again which were sent to the state lab. We would be following up with a specialist in Boston at Children's Hospital on Monday (Dr. Marsden) to discuss the situation. We would

not be able to get the results for 4 or 5 days which seemed like an eternity. We arrived home Saturday night and we were all very scared and sad. I had never really knew what the new born tests were for since there were so many rare disorders listed. I just always had the girls tested for everything. I was so afraid and also very ignorant to her potential condition which was also very rare (only 50 cases world wide). That scared me tremendously! I wondered if she would get the proper treatment.

Around 6:00 PM I received a call from our Pediatrician who apologized for the way the news was given to us and also explained a little bit more about her condition. She said although this is rare it's very well understood and treatable. This made me feel a little bit better. She reassured us that with proper treatment she would grow up normally, but our Specialist would need to explain all the details on Monday.

First thing Monday morning we called our specialist and were off to Boston. We met with Dr. Deborah Marsden, who would soon become our guardian angel through this whole ordeal. Dr. Marsden performed a series of tests on Olivia, including a urinalysis for organic acids and a skin biopsy for enzyme activity. She told us Olivia may have Ketone Utilization Disorder also called Beta Ketothiolase Deficiency. She explained to us everything that we would need to know and answered endless questions for my husband Dan and I. She explained that Olivia had an enzyme deficiency which inhibits her body from breaking down an amino acid called Isoluecine and if your body doesn't break down and excrete this amino acid which is found in all food with protein, toxic levels of acid from the protein will build up in her blood and she can have severe effects from this which could begin with vomiting, diarrhea, trouble breathing, seizure, coma or worse. Another issue is if she goes too long with out food more than 12 hrs a night or with inadequate calories during the day it will also cause her to have a metabolic crisis as described above. All of these terms were so new to me and I tried so hard to absorb everything she was telling us.

What I left with was that she needed to be on a diet (OK I can handle that), she can't go too long with out eating (I can handle that too) and when she is sick even with a cold her body may have a reaction as a result of the body fighting off the virus or fever (This one is tough). I wanted to put her in a bubble from that point on. Every child is going to become sick sooner or later, but if Olivia gets sick and won't eat we will have to go to Children's Hospital to put her on a glucose 10% IV which will prevent her body from breaking down it's own protein stores and prevent a crisis from happening. So basically this means she will be in and out of the hospital for preventative measures. A part of me was in such denial and confused by this whole thing. We went home and waited a few days for Dr. Marsden to give us the definitive results which came on Friday of that week. When she called it was around 5:00 at night and all week I anxiously awaited her call. I prayed that maybe this was a false positive and Olivia would be fine. I was wrong! Doctor Marsden called to give me the results and again her urinalysis was positive. I was so saddened, depressed and scared. The one thing I remember Dr. Marsden saying which did comfort me was "that it's a relatively benign disorder that is treated mainly with diet and that I still have a perfect healthy little girl". I hung on to that hope and those words stayed with me!

I held Olivia as tightly in my arms I dropped to my knees and wept. While I knew she was going to survive, I was so afraid for her and all that I know she will have to endure as she copes with this disorder.

When Olivia was 3 weeks old we went to our vacation home in NH and she came down with her first illness, RSV. We ended up leaving early to head to Boston to get her checked since I was concerned about her terrible cough. Once the Doctor evaluated her, they discovered that she had RSV Bronchiolitis and would need to be admitted to prevent any complications. She was in the hospital for 4 days on IV and boy was that draining. Seeing out little girl so sick with IV fluids and this very scary underlying condition was too much for me. I became very sad for a long time. I tried so hard to find a way to cope with this new life and challenges that my family would be faced with. I began researching over the internet and found that to be even worse since all that seemed to be available was the worst case scenarios. After reading everything I could out there I became even

more worried. I stumbled across a web site for children with Organic Acid disorders which had family stories and advice etc. for families affected with this disorder. This was helpful at first, but I soon discovered that for me hearing about other peoples pain was not allowing me to deal with my own. I then began to read about the positive ways people deal with different tragedies in their life and decided that I was going to do just that, focus on the positive. I realized that I have all the details I needed about Olivias disorder and a wonderful Doctor and best of all Olivia is doing excellent! Having said that I decided now I was lucky! Lucky that she was tested for this, unlike many children who were not. Lucky that she is alive and the final piece to our family puzzle is here. Lucky to have her and hopefully help others by advocating Expanded NewBorn Screening Tests and sharing our story. Lucky beyond imagination!

When Olivia was a few months old we noticed how chunky and adorable she was and began to nickname her OOMPA. From that point on everyone in our family has referred to her as OOMPA!

When Olivia was just over 1 year she caught a virus and was vomiting so we brought her to the hospital twice within one week to prevent a metabolic issue and returned home both times with in a few hours of her being on an IV. She had never become acidotic or had a crisis as of yet. Later that same week she started to lose her appetite and acted a little out of sorts. I could barely get her to eat 16 ounces of formula and no solid food all day. With the guidance of the on call Doctor in Boston, we kept a close eye on her managing to get her to eat little bits of formula through the night. Enough so we thought to keep her stable or so we thought. Then on Sunday morning things changed. When I went to her crib at 5AM to check on her as I was doing constantly throughout the night, I noticed she was breathing rapidly, and seemed very groggy. I put on her favorite TV show and she would still smile and rock back and forth to her favorite music. However her breathing concerned me and felt as though she needed to be seen. I called the Doctor on call in Boston and they advised us to see her local pediatrician to get a final answer on her breathing and lack of appetite. He checked her over and concluded that she most likely had become acidotic from not eating enough and battling a virus during the week and said we would need to go to Children's hospital right away! So we packed a bag and headed to Boston for the third time that week. On the way there Olivia became very lethargic and her breathing worsened. She was in and out of consciousness as if she was in a deep sleep. It was such a indescribable feeling to see your child lifeless and weak and not know exactly what was going on inside her little body. I knew something was desperately wrong! With my husband Dan behind the wheel, he was able to get us to the hospital faster than the ambulance ride we took with her earlier that week. It's amazing what you will do when your child is in need. When we arrived at the hospital Olivia's labs were drawn and the same Doctor from the ER that treated her earlier that week said she was a much different child than he had seen just two days earlier. Her bicarbonate level which was normal on Friday and at 22 had dropped to 6. This meant that Olivia had severe metabolic acidosis and would need a glucose IV ASAP! As we watched and waited for her to recover we had many concerns. She was still very lethargic and breathing very rapidly and not her usual happy self. She laid limp in my arms moaning with discomfort. It took almost a full 24 hours before she seemed to get better and her rapid breathing subsided. Her labs were checked every four hours and gradually improved and within two days she was discharged and made a FULL recovery. Thank god, my prayers were answered. What a trooper this little girl was, yet again proving to us that she was going to beat this! I recall sitting in the hospital room looking out the window which overlooked the city of Boston, thinking how quickly my life has changed and how fragile life is. This is my new path I was going to be traveling and one that would change me forever!

Many months have since passed and while she has had colds and ear infections, since then Olivia has done fantastic and had no other metabolic issues or hospital stays. We have allowed her to finally sleep through the night after a year of feeding her as if she was a newborn every 4 hrs so her body would not go in to a fasting mode where she would begin to break down her bodies own fat and protein which is essentially like eating a steak to her.

Olivia is approaching her 2nd birthday and has had to be in the hospital two more times (once in October and

then a second time on Christmas) for viral infections as preventative measures so we never get to that point again where she is in severe crisis. Once was enough for us to know her limits.

Olivia is now 3 ½ and doing better than ever! She has not been in the hospital in a year, thankfully! It has most definitely gotten easier as she gets older and can communicate to me how she feels and also understand the importance of her eating and drinking. I have found many little tricks to get her to drink and eat when sick so that she doesn't end up in the hospital. This has given me such a relief to see her doing so well and less and less hospitalizations! She is still so small yet I feel like she understands her body already just by the foods she chooses to eat. She is truly a miracle from God as are her big sisters who love her more than anything! They adore Olivia so much and make each day such a pleasure for me and Dan!

We will celebrate her 4th birthday in February with our family and what a celebration it will be! She has so much love and support and some day when she is grown, she will appreciate all that she has.

She continues to do so well and is saying everything. She is just perfect and such a gift! She is thriving like all three year olds should be and hitting and exceeding all of her milestones both physically and developmentally!

She makes us laugh so much and smile all day! She is a little genius! She is loved by her sisters, parents, extended family and friends and medical staff who we are so blessed to have. They have truly been there for us every time we have needed. Without them I don't know where we would be. She is such a little miracle and a constant reminder of how blessed we are.

It's amazing how life throws you a curve ball and yet you still find a way to catch it! I have learned more about myself and what life is all about in the past three years of my life than I have in the prior 33 years. I will never take one minute for granted and will always praise the lord for everything I have. I truly live one day at a time and enjoy life no matter what the day may bring. My family is so amazing to us and my adorable children and my husband who have all been such a pillar of strength for me!

I will continue to learn as much as possible about Olivia's disorder and how it affects her, but I feel empowered by all that I have learned so far. I will continue to ask questions and learn as much as we can, as well as do all that I can to help raise awareness for ENBS and her disorder.

I am sure we have many challenges ahead of us as does everyone in this world, but one thing is for sure, I know now that I can face them and grow from them and having weathered a storm I know brighter days do come! Life is a gift that we have all been given, no matter what the journey may be and Olivia has allowed me to realize and embrace this gift and to appreciate every moment that we have!