

# Caitlin's Story

March 2012

Caitlin is our 3rd child. She was born a year after our 2nd child Nicole, who passed away from severe heart problem. On the day Caitlin was born, I held her in my arms and cried with joy and was thankful for having a healthy "normal" baby girl.

The first 4 years of her life were uneventful, though I've noticed changes that were questionable. I noticed that when she was around 3-4 years old, she would have occasional incidents of tripping/stumbling and questioned myself why is this happening but I blamed it on her footwear not thinking it is a sign of muscle weakness.

When we would go to the mall she would complain of being tired after 30 minutes of walking and would ask to be carried. I thought she was just being lazy like any typical toddlers.

She started having bouts of diarrhea and abdominal pain that comes and goes with no definite diagnosis from her doctor but mainly assumptions that it was a stomach flu or from oily, fatty foods.



Caitlin born September 2, 2003  
Weight 7 lbs 2 oz.  
Length 19 3/4 inches.



August 18, 2008,  
Pre-Kinder First day of school

She is skinny in spite of having a great appetite, her weight gain was 15% of the growth chart which was assumed normal for her since being skinny runs in the family.

It was in August 2008 when everything started. First day of school, Pre-K, she got sick with high grade fever and vomiting and diagnosed with Urinary Tract Infection. She got treated with antibiotics and the bouts of diarrhea and abdominal pain went away. After a few months, she started having bouts of diarrhea again and found out from previous laboratory results that her liver enzymes were elevated. A repeat liver enzyme test and celiac disease test revealed she has celiac disease.

After consulting with Dr. Jatla, pediatric GI in Temple Texas, test showed negative for Celiac disease and normal endoscopy and gastroscopy test. The liver enzymes remained elevated, after multiple blood test and no definite answer, a biopsy of the liver and muscle were done. The biopsy showed high levels of glycogen thus the diagnosis of POMPE.

After I found out what I was dealing with, I went on full gear and made appointments to all the doctors that she needed to see, tests that needed to be done and contacted organizations that can help me and joined support groups.

I changed her diet to high protein, low carbohydrates and added a can of pediasure 2-3 times a day. When she was diagnosed in March 2009 she weighed 31 lbs.

Caitlin started her Myozyme treatment in July 17, 2009 at Dell Children's Hospital in Austin Texas under the care of Dr. Gibson. She had a port put in after having several problems with cannulation. It was emotionally and physically draining especially for Caitlin. She had treatment for 6 months in Austin Texas and now goes to an infusion center in San Antonio Texas.

Caitlin turned 8 last September 2, 2011 and went to see Dr. Gibson. We marvel on Caitlin's improvements since she started her Myozyme treatment. Now, she's 4 ft tall and weighs 49 lbs, her upper and lower extremity muscle strength has improved tremendously. She's able to stand on one foot on her own without wobbling. Caitlin is able to play jump rope by herself. When going upstairs she does not hold on to the rail for support or crawl or ask to be carried. Getting in the car, she does not need to be pushed anymore. She can jump into the bed or exam tables without help. It is easier for her to stand up from a sitting position. She walks longer distances without easily being tired or complain of leg pain. Her waddle walk has improved. Most importantly, her lungs have improved, last year she only got sick twice from upper respiratory problems.



Caitlin at aged 8, taken October 2011 at the AMDA/IPA Pompe Conference in Texas.

All these improvements has given us especially my daughter great joy since she does not feel left out, she can enjoy being a kid even though she has limitations.

**By Ruby Naldoza  
Caitlin's Mother,  
USA**



July 17, 2009. First day of ERT treatment in Austin, Texas



AMDA Fundraising, April 2011, Texas



Caitlin playing Flute as part of her Lung exercise