

# “Live with Morquio”

## Story 1

Written by Stephanie Bozarth,

### 1. Birth: healthy baby!

Annabelle was born on May 30, 2006 at 10 lbs 8 ounces via c-section. Due to her large size, that was the recommendation by the medical team. Although a large baby, she was perfect in every way. She appeared healthy, strong, and vivacious.



5 days old

A healthy beautiful baby with no sign and symptom!!

### 2. Infant: Initial notice!

During Annabelle's first couple of months, I began to **notice a curve in her back** when I held her to my shoulder for burping and cuddling. I attributed in my mind to her large size in utero. At her four-month well baby check up, I told the pediatrician my concern about the curve in her vertebra. The pediatrician was not able to detect the curve while Annabelle was lying down however it was apparent when held up to her shoulder. Our pediatrician gave us a note for x-rays for further examination. The following day, I took Annabelle for x-rays at the local children's hospital which did confirm **four anomalies in her bones to suggest a metabolic syndrome.**

### 3. Diagnosis: Worry and Surprise!

Within days of the initial x-rays, the doctors suggested that Annabelle may have a metabolic syndrome. We had spoken with our pediatrician and had a genetic appointment set for the next week. We were told initially that her diagnosis may be very serious and it would be a process to rule out various syndromes. The whole diagnosis process of blood work, urine analysis, and skin biopsy required two months to complete the diagnosis. By 6 months of age, the diagnosis was confirmed as Mucopolysaccharidosis IVA (MPS IVA: Morquio A Syndrome). Throughout that process, we spoke frequently with our geneticist at appointments and over the phone. I felt informed and involved with the process.



7 months old



7 months old

She had a slight bump in her spine which we noticed at 4 months old.

#### **4. Community with other Morquio families**

Austin and I connected with the Morquio community through intensive online researching of Morquio Syndrome. We quickly found the yahoo group for those affected by Morquio syndrome. As we veraciously looked for more information, treatments, cures for Morquio syndrome, we developed relationships with other families in the Morquio community. When Annabelle was 8 months old, we attended our first Morquio Syndrome fundraiser for a child in New Jersey. We had to travel far for the fundraiser but it was every bit worth the effort. We met other Morquio families and children along with Dr. Tomatsu. Connecting via internet is one way to connect but actually meeting face to face was very worthwhile. Since Annabelle's diagnosis, we have continued to make friends and connect with Morquio families around the world. The support from other parents is a life line of hope to get through some of the tougher times dealing with Morquio. I have connected closely with parents of children close to Annabelle's age. Because she was diagnosed unusually early, we had more time to digest the information and create a network prior to any major medically necessary surgical interventions.



**1 year old**



**1.5 year old**

## 5. After diagnosis, progression of the disease

Annabelle's disease is progressive. Over her short four years we have watched the changes in her bones through regular x-rays, the **changes in her gait, the way she tires more quickly than her peers, and we see her peers growing heads above her.** When she was a toddler, there was nothing obvious to demonstrate that she had a syndrome. As she gets older, it is more and more apparent to our friends, family, and strangers. Sometimes strangers take a double look at her. I always smile in return and if they inquire, I take it as an opportunity to educate and spread awareness about Morquio syndrome. Every interaction is an opportunity to spread our network of friends in the community.

## 6. Experience of surgical procedure

Annabelle had ear tubes placed at 8 months old due to conductive hearing loss and repeat ear infections. At **2 years old**, Annabelle had cervical cord impingement which was initially diagnosed through x-rays and MRI by Dr. Mackenzie at Nemours Alfred I. DuPont Hospital for Children. She was prescribed to wear a neck collar to protect her from injury and given a surgical appointment. During our 3 months we waited for her surgical date, I found Annabelle screaming in her bed that bugs were all over her. She did not have bugs on her. She was experiencing tingling from the cord impingement. **Soon after, she had a spinal cord decompression and fusion surgery with Dr Mackenzie at Nemours Alfred I. DuPont Hospital for Children.** The recovery required her to wear a medical halo for 3 months. Although the halo was quite overwhelming, Annabelle adjusted within weeks to the halo and was up and about engaging in normal 2 year old activities.

Annabelle's second major surgery was hip reconstruction and bilateral knee plates again with Dr Mackenzie at **4 years old**. This was a two-part surgery which started with the left side reconstruction and knee plates and one week later she had the right side reconstructed. She was in the hospital for almost 2 weeks. The recovery was 6 weeks in a **spica cast**. This is a full body cast from the chest down to each ankle. The spica cast was defiantly the hardest recovery for Annabelle. She experienced persistent itchiness for the entire six weeks. There was a lot of pain in the first few weeks however that did subside over time. The spica cast was also very limiting in what Annabelle could do to feel independent. She was frustrated, scared, and anxious throughout most of the six weeks. The only times she seemed at peace was during visits from friends and her reflexology appointments. The key to this recovery was keeping her as distracted as much as possible from the spica cast.

Both surgical interventions went very smoothly with Dr Mackenzie and his medical team. I attribute Annabelle's amazing recoveries and zero complications to making sure we had the best orthopedic doctor experienced in spinal dysplasia syndromes. We were also confident in the anesthesiology team at Nemours Alfred I. DuPont Hospital for Children as they too are very experienced in working with children like Annabelle and understood all the risks involved. As Dr. Muenzer, a prominent MPS researcher from UNC, once told us, finding a good Anesthesiologist is more important

than the surgeon as the most significant issues usually arise from complications during this aspect of the surgery. Finding competent and experience medical care is one of the most important pieces to caring for a child like Annabelle.



**2.5 years old**



**4 years old**

**After cervical fusion surgery at 2.5 years old, Annabelle is wearing halo ring (left picture).**

After hip reconstruction and bilateral knee plates at 4.0 years old, Annabelle was in a spica cast for 6 weeks. **(Right picture).**

## **7. The current situation**

Annabelle is currently still recovering from the hip reconstruction surgery with **Physical therapy on both land and water 5 days a week.** The first couple of weeks of recovery were difficult as Annabelle's muscles had atrophied, however since then, she continues to make great improvement every day in walking and endurance. Annabelle attends preschool 3 days a week and is enrolled in the public school Child Find program that ensures that she is properly accommodated in the learning environment despite any physical limitation. The Child Find Program is mandated by law to support children in the learning environment that may have any type of disability or limitation. We are happy that she is already in the program so that her transition to kindergarten will hopefully be smooth.

Now that her neck has been stabilized, hips reconstructed, and her knee plates will slowly allow her knock knees to self-correct over time, we don't expect any more major surgical interventions for a while. We continue to have biannual appointments with pediatrician, ENT, audiologist, ophthalmologist, cardiologist, and her orthopedic surgeon Dr. Mackenzie to monitor her condition. We know that severe Morquio patients tend to have cardiology issues in their 20's.

Annabelle was recently diagnosed with mild to moderate hearing loss in both ears which is common in severe Morquio A Syndrome. We are in the process of getting her hearing aids. The ear molds will need new ear impression and mold replacements made every six months and her hearing aids need to be replaced every three years.

Annabelle will also need to incorporate a pulmonologist in to her care in the future. However at this time she does not need it.



**4 years old**

#### **8. What do we expect from now on our kid?**

My expectations for Annabelle do require a balancing act of recognizing the behaviors of a 4 year old with need for structure along with consideration for her special needs that may cause more frustration. I want my daughter to grow up as a confident, happy child. I do expect her to participate in cleaning up her toys and personal hygiene as any 4 year old should learn. However, I do recognize that some days may be tougher than others if she is not feeling well or overtired. Overall, I hold high expectations for my daughter and gently encourage her to reach her full potential in everything.

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