# Cari

Cari, mother of Dylan (age 11) and Jessie.
Jessie had Sanfilippo Syndrome and died at age 13.





Cari: By the time she was one, things started to be going a little bit slower in the progression and so luckily I had a great pediatrician who really listened to me and thought that we should get early intervention. So we had her evaluated for early intervention and she started receiving services. Her progression picked up and she started doing really amazingly well, so we weren't concerned after that for about six months. At 18 months, her gross motor skills were really falling behind her peers and again we went to our pediatrician and he referred us to some physical therapy because she had some very tight heel cords. And so, for about six months, we tried different interventions to help with her heel cords, we tried serial casting, we tried botox injections and nothing really helped the tightness in her heel cords. So our pediatrician finally thought, well, maybe there is a neurological reason why her heel cords were so tight. He said "I've never seen heel cords this tight before" and so he referred us to a pediatric neurologist. When she was two, we went and saw the neurologist and she evaluated Jessie and thought that yeah, she's behind, and so we did a bunch of testing for some very common genetic conditions and those all came back negative, and so she thought that maybe Jessie had some mild form of CP. My husband and I went home and we talked about it and we thought, okay, this is something we can deal with, she might have some difficulties but we can deal with CP. We had a brain MRI done right before she was about two and a half and it came back significantly abnormal and actually the radiologist had recommended testing for a Mucopolysaccharidoses or MPS. That was the first time I had heard of MPS and so we didn't know what type and we went home and of course I read on the internet all about MPS and the different types and I knew as soon as I read Sanfilippo Syndrome, I knew that's what she had.

Cari: So, Jessie had a lot of energy when she was a toddler and that was one of the symptoms of Sanfilippo Syndrome. She was also very social, she loved her books, she loved videos and these were all symptoms that I read about for Sanfilippo. And then also the delays, the delay in language development, the gross motor delays, they fit well. Once a doctor had told me about MPS and I read about all the different types of MPS, her symptoms fit best with Sanfilippo Syndrome instead of the other types. So then the neurologist, had us come in she said yes, I have you in here to confirm that it is MPS3 Sanfillipo Syndrome. She told us that these were going to

be the best years of Jessie's life. That we were leaving and not to read too much about what was going to come next, but just to go home and enjoy her and there was really nothing we could do other than as she started to have symptoms later in life, we could try to support her through that, but there was no treatment or cure or anything really we could do at that point.

# What does this mean for having other children?

https://youtu.be/I6uyDXJJeEc

Cari: So when we got the diagnosis and we learned that it was a genetic condition, I think that I wasn't able to start the morning right away, because I was so panicked about my new pregnancy. I was only about seven weeks pregnant. I had just found out I was pregnant with our second child and then when Jessie was diagnosed, I found out that there is a 25 percent chance that this other child will also have Sanfilippo Syndrome. So, I feel like that inhibited my natural grieving process for Jessie because I was so concerned with the new pregnancy. So, that added an additional wrinkle for Matt and I, and we were really focused on what we were going to do about the pregnancy before we could even think about what was going to happen to our daughter. The next day, we started thinking about what we're going to do about the pregnancy.

Luckily I had a very good pediatrician who was so helpful and really anytime I called his office his secretary knew, and they would get him out from a doctor's appointment with another child, and he would drop everything and get on the phone with me and kind of help talk me through what we could do, what our options were, what we wanted to do and thinking about having another child and what that meant for our family.

Cari: So, I think that our pediatrician helped me to take a step back and he encouraged me to wait and get the Amnio, get the results and not to get too far ahead in my thinking. Of course the day after Jessie was diagnosed, I wanted to know immediately what is going to happen with our family, are we going to have another baby, are we just going to have Jessie? But my pediatrician helped me relax and take things a little bit slower day by day because I did have to wait until I was far enough along in the pregnancy to have the Amnio and then I had to wait for the results to come back and that took a long time.



## A LONG INDEFINITE TIMELINE

Your day-to-day life can be a happy place even though your child is going to die.

https://youtu.be/Rhn8BJeH6NU

**Cari:** When Jessie was diagnosed, I didn't know that you could become comfortable with the knowledge that your child wouldn't be there forever, and that you could still have a pretty happy

cohesive family with the child that has a terminal disease. We didn't realize that until we were living it but it's true, I mean we did a lot of things with Jessie and obviously there were some things we couldn't do as a family but we found other things that we could. We are better off having had her with us for the time that we did as a family, she really gave us such a gift and it makes it such a loss to have this huge hole in our family because she was just a big part of our family in all aspects, such a presence. So, I want -- I would like parents to know that your day-to-day life can become a pretty happy place even though you're facing the fact that your child is going to die at some point. That on a day-to-day basis you can have a happy family and not be focused on that all the time.



Cari: So, when we made the decision to take Jessie to Duke for treatment, we knew that it wasn't going to be a cure, they were very honest with us, we had hoped that it would prolong her life so that she would live longer but our ultimate goal was her quality of life and we just wanted her to be happy and to suffer less with Sanfilippo. Definitely we were thinking about if this prolonged her life by say, 10 years so she lived 20 years instead of dying early in her teens that that would give more opportunity for a cure to be available and that it would save her eventually, that there would be a subsequent treatment that would then be a cure. I think that at the time that we went to Duke, there was nothing on the horizon. There was nothing eminent and then several years after she had the treatment, there were some other gene therapies or enzyme replacement therapies coming down the line but things were moving very slowly. We did one fundraiser, we had a jog for Jessie in our town and it was great and we raised money for that research and then we just found that as a family the fundraising was really not one of our strengths. We had a very difficult time with fundraising and so we only did that one fundraiser but we were hopeful that cure would be in time to save her.

Cari: So, Jessie's progression was so slow that we remained hopeful for quite a while that she would live to see a cure for Sanfilippo. I would say it wasn't until maybe a year before she passed that she started to decline and our hope began to diminish. I wouldn't say that we were out of hope but it started to look like she wasn't going to live as long as we had hoped. I mean that she wasn't going to see a cure. But I don't know, I just -- I guess it was something that in the back of our minds we were hopeful for very long time, maybe we were naïve, I don't know, but she really did very well for a child with Sanfilippo and she had a very short decline which isn't common. So it gave us more time to hope that she was going to continue on this slow decline. When she was first diagnosed, the doctor told us she'd probably live 10 years from diagnosis. So, she would be gone at 12, and that was just really a hard pill to swallow. And I didn't accept it and then she did pass at 13 years old.



Cari: Our pediatrician helped guide us to different specialties that would pop up as Jessie progressed in her disease and had different symptoms. She had a GI doctor, and a neurologist and so I really assembled what I thought was the dream team. We didn't just go to children's, and when a doctor that we were seeing left childrens to say go to somewhere else we followed him to talk. We had specialists all over the city but it was really our pediatrician that coordinated her care and I am very thankful that he has a whole child perspective. He didn't see Jessie as just a child with Sanfilippo. He first saw her as a little girl, a child and that know she did have Sanfilippo, but he continued to see her for well, doctor checkups and child visits. And he was our first go-to when something would come up and then he would help to refer us to other doctors. So, really, he was our biggest support.



### **DECISION-MAKING AND PALLIATIVE CARE**

Once we understood what it was, we got on board pretty quickly. https://youtu.be/L1gmUku5uec

Cari: During one of our visits, the emergency room doctor had seen that we had been in before - just a few weeks before. She asks me if we had taken advantage of their palliative care team and I said no, but it was something that I had thought about and I was very interested in. Just in terms of managing her pain or discomfort because I had a very low tolerance for seeing her in pain and same with my husband Matt, but when I did, suggested to Matt, he right away was very upset that I was thinking this is the end of life. That's not what I was thinking at all, and so it took a little bit of talking and he visited their website and he did agree to meet with them and they did an amazing job of explaining that it wasn't for end of life care, although, you know several months later, that's where we were. But yeah, we got on board pretty quickly.



### **SPIRITUALITY**

The church provided emotional support for us, though my relationship with God has been affected.

https://youtu.be/V3WYIN-aMk8

**Cari:** So our family belongs to a church in Framingham, it's a congregational church, Edward's church and we picked that church because the head of Christian education had a special education background and when we went to visit as a family, Jessie was in a wheelchair and Dylan was in kindergarten and we were welcomed with open arms. They had a program that

included Jessie and so it was a great fit for our family and the congregation just took us in and they adored Jess. So it was a great place that we could do something as a family because when you have a child with a rare condition or a disability, sometimes it's hard to do things together as a family. It's something that we could do together every Sunday morning. So, as Jessie started to decline, the church provided a lot of emotional support for all of us. It was traumatic for them too because they love Jessie, but they were there to support us and help us through her decline and passing.

**Cari:** I don't have a very strong relationship with God at this point in my life. We still go to church and I really appreciate the community and the spiritual part aspect but not necessarily my individual relationship with God. I'm not sure where that stands, but I want to support my husband's relationship that he has with God and my sons. So, I feel like I'm just going through emotions right now, but that's what I have to do.



#### **SIBLINGS**

Her sharp decline was hardest on her brother. He thought he had more time. https://youtu.be/AsOSDRS4E7k

Cari: When he realized that Jessie wasn't going to have a full life and that she was going to die before him it was hard. We always gave him the optimistic view that we didn't know how long she was going to live and that she may live to be 30 or she may live to be 20. He, a few times, had said if you and dad die, I'll just take care of Jessie, she can just live with me. So, he was well-prepared for her to live very long time, and that was why her sharp decline was the hardest on Dylan, he was not prepared for that, he thought it was going to look a lot different. He thought he was going to have years of getting used to the fact that she was going to be gone and it was really sad for him when she stopped being engaged with him because she was in so much pain. You know we reached the point where if she was awake, she was uncomfortable and when we gave her the medication, she would be pretty much knocked out, and so there was a brief period there where she would be awake and she wouldn't be able to be engaged with Dylan. The day after she died, he said that one of the things that he really regretted is that he didn't get to say goodbye to her which is just crazy because he said goodbye to her so many times, the week that she spent in our home before she died, but in his mind, he wanted to say goodbye to her when she was still engaged with him and that she would hear it from him. So that was really hard and that can't happen, how could that have happened, that we had realized when she was still engaging with us and happy that she was going to be gone in a few weeks and so now say goodbye to her. I mean it's just impossible to have that point, it doesn't happen. But when you're 10, that's what you want and so, he will tell you that's his one regret.



Cari: So, I think that through this whole experience with the family that Matt and I get the most support from each other, and it's really our marriage that has supported both of us through this horrible experience although we both always laughed that, you know we couldn't have done this without Jessie. She was like a major support during her life because she was so happy and in the moment and helped us deal with it. And certainly, my son too, you know, children have such a great way of living today like that's the only important thing. And it helps when you have a child with a terminal illness, it really helps to live in the day and so your children help you to do that because I certainly wasn't like that growing up and as an adult, I was very goal oriented and planned ahead and had to know what our five-year plan was and then you have a child with Sanfilippo and that just goes all out the window.

Cari: I feel like Matt and I were lucky to always be on the same page. I mean we talked about everything. I mean we really talked a lot about where we saw things going, what was important to us in terms of Jessie and her life and her quality of life. And so, I feel like we were really lucky to have a relationship where we were both open to talking about whatever at anytime and so before it was clear that she was dying. We had been having that conversation for about 10 years. And so, that was really helpful and I know that not all couples are there but we were really lucky.