Barth Syndrome Foundation®

Family Focus

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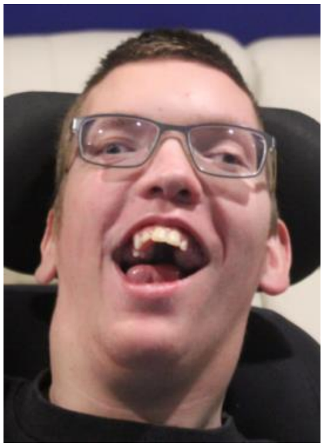
MEET JARED
Interview with Jared's mom (Susan)



How old is Jared Now?

He is twenty-six.

Jared both has cerebral palsy (CP) and Barth syndrome (BTHS). How has that been a challenge?

One of the biggest challenges has been in how much CP and BTHS overlaps. There is a lot of overlapping with regards to his muscle tone and his muscle wasting. Sometimes it's difficult to determine what is bothering him. The more we learn about BTHS, the more difficult it is to figure out what is bothering him. Is it the fatigue? Is he tired of sitting in his wheelchair twelve hours a day? Is he in pain? He can't tell us because he is nonverbal. He doesn't like lying down and he can't sit anywhere else. So, it's really challenging. Like others with BTHS he can be fine one minute and thirty minutes later he is extremely ill. When he gets ill, he can spike a fever of 104 degrees and start vomiting and struggling for breath. His major issue is in getting pneumonia. He doesn't get the mouth ulcers, but he can get very sick with a bacterial infection. He does get the pneumonia and flu vaccines.

He's tube fed and won't eat anything by mouth. I don't know why. Is it a BTHS thing? He has all the food sensitivities. It could be BTHS or it could be CP too. We have tried for twenty years to get him to eat but it just isn't going to happen. You would think you are killing him if you come at him with anything to put in his mouth. He doesn't like anyone touching his face. He is hypersensitive about that.

He communicates with us with his eyes, but he refuses to use the technology that we have introduced to him. I just don't know why.

Can he let you know when he hurts?

He will let us know when he is in pain. It is difficult to figure out why he is hurting. We can't tell if it is a muscle cramp or if his bones are aching as we have heard described by other people who have BTHS. He is generally so good natured. He is a very happy guy.

Do you have to physically lift him to adjust his position when he is like that?
He is too big for me to do that. His father is so good with him. We call his dad the master and commander of Jared. He has a system. Believe me if I get one step out of line, I hear about it.

Is he taking GCSF?
No. We have never done a neutropenia study with him. Sometimes his neutrophils are on the lower levels but most of the time his levels are normal. There have been times when he has had absolutely no neutrophils.

His older brother, Jordan also had Barth syndrome. At the time we didn't know that. But looking back, he was constantly neutropenic. He spent more time in the hospital than out of the hospital in his three years. But the year after his transplant before his death was a very good year. He died from overwhelming sepsis. He picked up some bug and that was it.

So, you didn't know Jordan had BTHS before Jared was born?
No. We asked the doctors if there was any way this could happen again. They told us it was just a fluke. We were told that we could have other children. I had another son, and everything was fine. Then I got pregnant with Jared. Around the sixth month of pregnancy, I wasn't gaining weight and he wasn't gaining weight. As soon as his lungs were developed enough, they induced labor. About a year after Jordan died his cardiologist sent me this article about BTHS and suspected that may have been his underlying diagnosis. I sent it to the doctors where he had his transplant. The doctors told us they didn't think he had BTHS. So, we didn't think about it again.

For some reason the article was placed in Jordan's file. When Jared began having complications, they pulled Jordan's file. The asked us if we had heard of BTHS. My heart sank. They were both born with hypoglycemia and Jared was in heart failure. They were both smaller babies.

They kept monitoring Jared's heart. It went up and down. For the first couple of months it went back and forth but to this day his heart function is fine.

He was born in 1994 and he had the genetic testing in 1996

Where did the CP come in?
When he was around four months-old he needed a hernia repair. Unfortunately, during prepping him for surgery he became very unstable. A code was called which you could

hear everywhere in the hospital I panicked but when no one came to get me, I figured it couldn't have been him. In my heart, I feared it was him. Then an hour later they came for me and I knew it was him.

I asked if he could have brain damage. I was assured that he wouldn't because he wasn't without oxygen that long. As the days went by It became clear they were wrong. It was horrific. No one would come right out and tell me your son has brain damage or your son has CP. It was a total reset. All of his milestones were lost.

We got him in to see a neurologist several months after he was discharged from the hospital. Two minutes after meeting this doctor he said "*I'm so sorry, your son has cerebral palsy. He is never going to walk, talk or go to school.*" I was floored. We had both taken so much time off of work with Jordan and now with Jared. I had no idea what I was going to do. Once they got his seizures under control, he was transformed into the happy guy he is now.

Ironically, he never had the hernia repair.

So, you received the diagnosis of BTHS for him after you learned he had CP? Wow talk about a double whammy.

It was not big deal. I knew in my heart he had it. It was just a confirmation. The internet was just coming of age around this time. We had been looking for information using this new tool. Eventually we did a search and found you on the first website you built.

Had you met other Canadian families?

No, we didn't meet anyone in person until that first conference in Baltimore back in 2000. We came without Jared for that first meeting. We met the other families from Canada, but we didn't really connect until the second conference. It was hard because all of the boys were alive and none of them were in wheelchairs like Jared. None of them had CP. I was trying to find answers from the doctors who were at the conference. We were asking the doctors if they knew of others with BTHS that had CP. We were told there weren't.

What do you remember about that first conference?

I remember standing next to Dr. Barth who seemed fascinated by seeing so many children in one room with BTHS. He and the other doctors were looking at how they would use their hands to get up from the floor or how they would walk and run. There was this glass window where people would just stand to watch all of them play.

Facebook has been great to keep up with each other but there is nothing like getting together and seeing the similarities. There are so many benefits to just watch and observe the little idiosyncrasies. We have that little window into BTHS when we are doing the luminaries in the sand. It is very hard for them to walk in the sand.

I met one of my best friends in the world through this organization. I can't imagine my life without her. I usually get to see her a few times a year. The worst part of this pandemic is that we didn't get to go to the conference, and I don't get to see my best friend.

Tell me about what it is like having daughters who might be carriers.

I have an immense sense of guilt that I passed it on to one of my daughters. Learning you are going to be a grandmother should be a time filled with joy. I remember having an overwhelming sense of dread wishing, "Please don't let it be a boy." But then you realize that a girl might be a carrier. The day we learned my daughter was a carrier, we both just broke down and cried. Then the day when I learned my other daughter wasn't a carrier, I cried but for altogether happier reasons. I was so relieved.

What is a day like with Jared?

We have a lot of routine with Jared. Prior to the pandemic we would get him up, dressed and fed and ready to go at 8:00 AM to attend a day program. He loves being around other people. He likes watching people interact. He doesn't need to be entertained. He just needs to be around people. He comes home around 3:00 PM. He gets a feed right after he gets home. At 7:30 we get him ready for bed and he goes to sleep.

Does he have any favorite?

He loves his nieces and nephew, but his sister Jennifer is his favorite person in the world. He loves all of his siblings, but he loves her the best. He loves his Talking Tom app which goes everywhere with him. He loves people to read to him. He liked television but right now he is over it.

Any final comments?

We have lots of good times. We don't exclude Jared from anything. He goes everywhere we go. He is a part of our family. He has taught us a lot of things, but I am not one of those people who say "*I wouldn't change a thing*" because damn it I would. Without hesitation, the one thing I wish I could change would be Barth syndrome. I would change it in a second if I could.