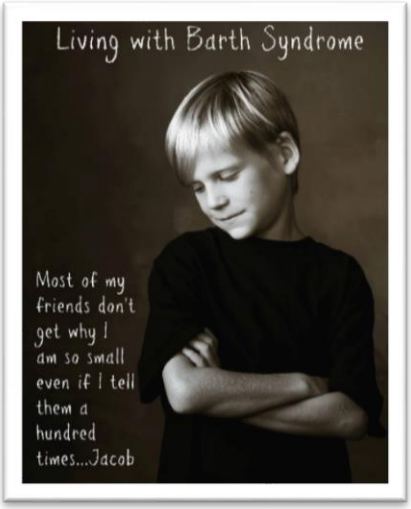
Barth Syndrome Foundation®

Featured Story: Meet Jacob



Jacob Anthony Wilson was born August 22, 2000 after a normal pregnancy. According to his mother, Amy Wilson, as an infant he had a normal appetite. He was behind on some things but nothing that would indicate that she or his physicians should be alarmed of.

"You just trust that the doctors would tell you if something was off, you know?" said Amy.

Jacob sat up later than other children his age and suffered from recurring ear infections. He had a hard time fighting them off and no one could explain why. Pulling up was more difficult for him due to overall weakness. Jacob started crawling around 8 months of age and began to walk at 14 months.

But nothing seemed particularly abnormal until a week after his first birthday. On August 27, 2001, Jacob suffered his first seizure. This was said to be a febrile seizure caused by virus. Jacob went on to have 5 seizures altogether.

On October 29, 2002, at the age of 2, he was hospitalized with an ear infection and low CBC (Complete Blood Count). This started a routine of weekly blood draws at the local children's cancer clinic located at University of Mississippi Medical Center's Blair E. Batson Hospital for Children in order to monitor the low CBC's. His mom, Amy, had lots of fear that something bad was going on that his doctors weren't finding. After 3 weeks Jacob's CBC came back up and his mother was just told that Jacob just had more trouble than others fighting off infections.

Jacob

In the meantime, Jacob also had complaints about leg pain. He had an x-ray of his legs done in 2004. He had coxa vara, a deformity of the hip, whereby the angle between the head and the shaft of the femur is reduced to less than 120 degrees. This results in the leg being shortened, and the development of a limp.

But the reason for his pain was said to be caused by rheumatoid arthritis. Jacob started treatment and physical therapy.

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(L-R) Damon, Jacob, Kristi & Marvin

His short stature was addressed in 2007 and endocrinology determined that he should be on growth hormones.

Rheumatology also decided to recommend Jacob go to a neurologist because of tight tendons. It was the neurologist who determined Jacob to have a myopathy (muscle weakness). Jacob had a muscle biopsy performed because of myopathy.

He now had many different individual diagnoses but no overall explanation.

Jacob was brought in to genetics after his first cousin Christopher's diagnosis of Barth syndrome. Myopathy, unexplained viruses, skeletal problems and questions with his CBCs in the past fit the same diagnosis. Geneticist Omar Abdul- Rahman was now on the case to find the cause of muscle weakness which was apparently affecting boys in the family.

Barth syndrome (BTHS) is a rare, sex-linked genetic disorder of lipid metabolism that affects males. Typically, boys with BTHS present with hypotonia (low muscle tone) and dilated cardiomyopathy (labored breathing, poor appetite, and/or slow weight gain) at or within the first few months after birth. Other important features of BTHS include bacterial infections because of neutropenia (a reduction in the number of white blood cells called neutrophils), muscle weakness, fatigue, and short stature. Although most children with Barth syndrome manifest all of these characteristics, some have only one or two of these abnormalities and, as a result, often are given incorrect diagnoses. It is very rare, and there are less than 200 known cases worldwide. It has been described to be much like finding a needle in a haystack for the doctors to discover someone to have Barth syndrome.

Lab work confirmed Jacob to also have the same diagnosis, making him and his cousin, Christopher, the only two known cases of Barth syndrome in the state of Mississippi. Now all of the questions and unexplained things happening to Jacob had a reason.

Kidney stones have been the main focus and arthritis to blame for being clumsy until now.

Today at the age of 16, Jacob is heart healthy, but in 2010 he had several episodes of arrhythmia. Thankfully he still does not present with cardiomyopathy, one of the common characteristics of Barth syndrome. However, another of Jacob's struggles are that several bouts of hypoglycemia landed him up in the emergency room. Boys with Barth syndrome are very high risk for hypoglycemia if they haven't eaten.

He has also recently had neutropenia rear its ugly head, causing him to miss a lot of school. Neutropenia is a reduction in neutrophils, a type of white blood cell that is most important for fighting bacterial infections. Neutropenia may predispose an individual to mouth ulcers, fevers and bacterial infections such as bacterial pneumonia and skin abscesses.

Jacob doesn't eat sweets but he craves sour and hot foods. He will eat as much hot sauce as he is allowed. It is much more difficult for him to manage buttons on clothing and keep up with his peers physically.

Upon asking Jacob to share some physical and social challenges of having Barth syndrome, his reply was, "I cannot walk that far without my lungs expanding and crushing my ribs and making me tired and drowsy. I cannot hold heavy objects up for very long or carry them very far. Most of my friends don't get why I'm so small even if I tell them a hundred times, but some of my friends get it, like my friend Michael. He has been there for me for as long as I remember and I appreciate that from him. He believes in me. But because of my size some kids try to take advantage of me and tell me what to do."

There is no specific treatment for Barth syndrome, but each of the individual problems can be successfully controlled.

Written by Kristi, Jacob's aunt and mom to Christopher, age 8, BTHS