



Barth Syndrome  
Foundation

#### Quote of the Day:

**"A central repository for clinical data provides a valuable resource for researchers. ... Only with a critical number of patients is it possible to know what is common and what is not, what is expected and what is not, and what works and what does not."** ~ *Gerald Cox, MD, PhD, Clinical Genetics, Children's Hospital, Boston, MA; Clinical Research, Genzyme Corporation, Cambridge, MA; Scientific and Medical Advisory Board, Barth Syndrome Foundation*

## Featured Story: A Personal Message from Michelle



Michael Anthony, Jr. & Michelle

The Barth Syndrome Foundation (BSF) helped save the life of my first born son, Michael Anthony, Jr., and my sanity. We would have been lost without the wonderful people that are a part of the foundation and the many doctors who support the foundation with their research and work.

My oldest son, Michael Anthony, Jr., was born on November 25, 2001 after a full-term, normal pregnancy. He appeared to be normal and healthy. We went home from the hospital two days later but had to return the same night to have him admitted for dehydration. He was nursed back to health and a week later we went home.

Michael seemed to progress normally for the next year and four months, although he was smaller than most children his size but his pediatrician did not seem concerned. She suggested we feed him more fatty foods to get his weight and size up. He did seem to get sick rather easily but nothing too severe. On March 30, 2003 everything changed.

Michael became ill and we took him to the pediatrician's office, which then sent us to the emergency room because he did not know what was wrong with him – but told us he was very ill. Michael was in heart failure and his absolute neutrophil count was zero. The nurses asked me if he had leukemia. He was admitted to Brackenridge Children's Hospital to the pediatric intensive care unit for three weeks, on which occasion he almost died twice.

Barth Syndrome Foundation  
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A week into his stay they decided to test Michael for Barth syndrome. His pediatrician did not believe he had this disease because there was no family history of it and it is a genetic disorder. The test came back positive and we had a diagnosis, which explained why he did not nurse well, why he was small for his age, why he was in heart failure, why his neutrophil count was so low, why he did not have a lot of energy, and why he did not eat well. All the pieces of the puzzle began to fit together.

We contacted the Barth Syndrome Foundation immediately. Within a few hours Shelley Bowen contacted us in our ICU room. She told us what to expect and where to find information. We went to the website and were able to pull documentation on the disorder and what could be dangerous for our son. We immediately printed the paperwork and passed it to the doctors, who then changed the treatment of my son and helped save his life. On April 20, 2003, Easter Sunday, we went home.



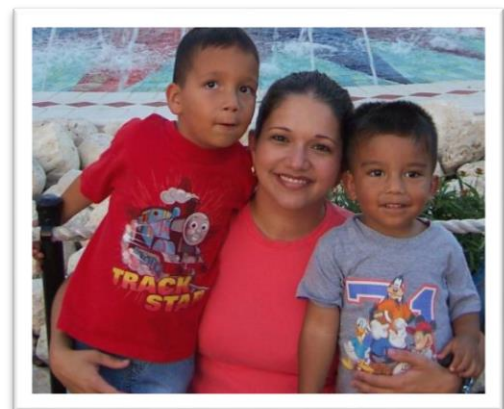
Boys and young men  
at BSF's 2008 conference

We joined the foundation and attended every conference from then forward. We learned all we could about Barth syndrome and managed Michael's health to the best of our ability. He began occupational, physical, and speech therapy and improved so much we were able to stop the appointments. He was on several heart medications and his heart function returned to normal. I would periodically give him neupogen shots when his neutrophil counts were low, but he never got a bacterial infection. Life seemed to return to normal.

I became pregnant with my second son in 2005. We were able to get prenatal testing with the help of the geneticists involved with the foundation and were able to determine before he was even born that he did not have Barth syndrome. Matthew Alexander was born on September 17, 2005, healthy and unaffected.

Michael began to attend public school when he was five. He was able to attend school all day every day and did not get sick often. It appeared to most people that Michael was not sick at all. Most people thought Michael had "gotten over" his Barth syndrome illness. Michael appeared to have a very mild case of Barth syndrome. His illness in 2003 seemed to be a blip on the radar and nothing more.

On April 18, 2009, Michael began not to feel well. The following day I took him to Dell Children's Hospital where he was admitted to the emergency room because he had developed a blood infection. Michael became septic and died on Monday April 20, 2009.



(L-R) Michael, Jr., Michelle & Matthew



Michael Anthony, Jr.  
(11/25/2001 – 04/20/2009)

I was in contact with the Barth Syndrome Foundation and the doctors associated with the foundation the entire time Michael was in the hospital. They gave me their love, their support, and their advice. A doctor with the foundation also spoke with the doctors treating Michael to help them better understand his illness and better treat him.

I don't know where I would be without the foundation. I was able to have six more precious and glorious years with my beautiful son because of the Barth Syndrome Foundation. They supported me through Michael's life, through his death, and continue to support me to this day. Several members of the foundation traveled from around the United States to attend Michael's funeral. I have received so much love and support from every member of the foundation.

I remain an active member of the foundation even though I have no living relatives affected with this disorder because this group has become my family, and I will fight in Memory of Michael and all of the boys and men we have lost to this terrible disease for a day when no one has to suffer or die from this dreadful disease.

There is no mild case of Barth syndrome!



(L-R) Michael Anthony, Jr.  
& his younger brother, Matthew



Michael Anthony, Jr. at his school's  
end-of-year Field Day (2008)