THE EVANOSKY
FOUNDATION

We Win in the End

A Father's Perspective

I think there are times in life when hardship and heartache occur and that a family's tragedy needs to be shared so that individuals and future generations can benefit. As a result, I find it important to tell you my story so that you have a solid understanding of my tragedy and take the opportunity to use the tools that you have available to affect change for the better. So with that, this is my story.

In November of 2001 my wife Sonya and I gave birth to twin boys, John and Christopher. It was a difficult pregnancy in that Sonya was bedridden for 36 days in the hospital and the boys were delivered early at 34 5/7 weeks by emergency cesarean. After the birth they both were in the NICU for 9 days due to a weak sucking reflex and difficulty regulating body heat. As far as their growth, they progressed normally, but were at the bottom of their adjusted age in reaching their developmental milestones. At 15 months, they hit a plateau and began physical therapy. At 24 months they were still unable to walk and we had the boys evaluated by a neurologist. John was given an MRI and it revealed damage to the white matter of the brain. The neurologist, using the facts that were presented--the premature labor, emergency cesarean, multiples pregnancy and NICU admission--deemed that the boys had Periventricular Leukomalacia (PVL), a form of cerebral palsy.

The boys continued to progress slowly and we had both boys involved in various types of therapy (speech, developmental, occupational and physical) to aid in their development. They were both developing nicely and could walk up to a quarter of a mile to the park in their Kaye rear walkers. We were setting ourselves up to live a life that we thought would include children with a mild case of cerebral palsy.

In January of 2004, Sonya and I had our third son, Jack. This pregnancy, opposed to the first, was a complete success. Jack was delivered by scheduled cesarean and in all accounts Jack was a healthy baby boy.

In the fall of 2004, we began to notice that John and Christopher were beginning to lose their milestones. They were losing the ability to walk, their speech was becoming debilitated, and their ability to eat was being affected. As a result, we had them reevaluated by the same neurologist who had made the original diagnosis of PVL. In December of 2004, at our insistence, he ran a metabolic panel of various tests. Sure enough, the outcome for both boys was that they possess the mutated gene that gave them the late infantile form of metachromatic leukodystrophy (MLD). Metachromatic leukodystrophy (MLD) is a progressive genetic metabolic disorder that affects the neurological system. Children who have this disease maintain their cognitive abilities but lose many of their developmental milestones by age two and are unable to walk, talk, or feed themselves. Life expectancy for the infantile form of this disease is approximately 6 years of age.

Upon this diagnosis, Sonya and I diligently researched what our options were for John and Christopher. We were told by members of the medical community that there was nothing we could do and that we needed to go home and make the boys comfortable. However, upon doing further research on the internet, we discovered that in some instances, depending on disease progression, a bone marrow/stem cell transplant was a potential option. We learned that Duke University was one of the premier universities in the world when it came to stem cell transplants and we immediately contacted a doctor there. Unfortunately, upon describing John and Christopher's physical symptoms, we were told that a transplant was not a viable alternative.

Once it was determined that John and Christopher had tested positive, Jack was tested the next week. Unfortunately, two weeks later, at 13 months of age, Jack also tested positive for MLD. Because he had no visible symptoms, the doctors at Duke indicated that Jack would most likely qualify for transplant. After a series of tests at Duke, Jack was deemed a viable stem cell transplant candidate and on April 1, 2005, Jack received an unrelated cord blood transplant.

As it currently stands, Jack is 15 months post transplant and is doing well. It appears that disease progression stopped somewhere between 9 and 12 months post-transplant. Because of his late diagnosis, there is some nerve damage to the peripheral nerves, but we hope through extensive therapy that Jack may be able to overcome the damage caused by an almost too late diagnosis. At this point in time, John and Christopher are still considered to be terminal but through very aggressive home care and the help of our extended family, their quality of life is excellent and they are two very happy little boys whose bodies just do not work.

Why is it important that I share my story with you? The purpose is to show you the importance of newborn screening for aggressive diseases that can no longer be treated after the symptoms appear. Early newborn screening provides parents with the opportunity to explore viable treatment options, develop care plans with the appropriate medical professionals, avoid the frustration of no diagnosis or misdiagnosis, and make appropriate family planning decisions.

Historically, newborn screening was only brought into play when there were treatment options available. Fortunately, technology has advanced so that some diseases may be diagnosed early, before symptoms appear. So be it MLD, or some disease that does not officially have a treatment process, it is important for the family to know about the disease sooner rather than later. In my particular circumstance there are a number of things that Sonya and I would have done differently had we known about the presence of MLD in our two older boys. First off, the boys would have been transplanted immediately after discovering that MLD was present. Duke University has had numerous successes with transplanting children early in life and that would have been an option that we would have thoroughly investigated. Transplant is a risky procedure and has roughly a 20% mortality rate associated with it, but untreated late infantile MLD has a 100% mortality rate.

Secondly, plans can be made to appropriately care for the child. Knowing that a child possesses a life threatening disease early in life certainly allows for treatment to be conducted right away, assuming that treatment is available. In the event treatment options are not available, there still is great benefit in knowing sooner than later. Both the practitioner and parent can begin to plan for the disease and in a sense stay one step ahead of the disease process. In doing so, the child benefits from this advanced planning and thereby maintains a higher quality of life than if the care providers were caring for the child but uncertain of the child's diagnosis. As NBS assays are developed for various diseases, both parents and practitioners will be in a position to make "best practice" choices for the child rather than reacting to what the disease chooses to do next.

In addition, many children that are affected by rare diseases often times are misdiagnosed or severely delayed in receiving a correct diagnosis due to the lack of experience of the diagnosing doctor. From personal experience, the neurologist who misdiagnosed John and Christopher was set up for failure from the start. My wife's premature labor and extensive hospitalization combined with my twins' premature birth and NICU visit predisposed the neurologist to thinking that the white matter damage he viewed on the MRI must be as a result of oxygen deprivation suffered sometime during this difficult pregnancy. In looking back, I think it would have taken a seasoned neurologist who possessed an "out of the box" thought process to look further into why there was damage to the brain rather than falling victim to obvious, misleading variables. By providing thorough NBS to babies, we take the potential clinical misdiagnosis by a licensed practitioner out of the equation. Unfortunately, my story in this particular area is a story often repeated throughout this country with many, many families being the victims of misdiagnosis.

In addition, had Sonya and I known of the presence of MLD in our family, we would have thought long and hard about having a third child. Even though stem cell transplant is a viable treatment for MLD, we would have thought carefully about putting a newborn through this if we had known in advance the challenges and risks of transplant. In Jack's 2 ½ years of life, he has suffered more than what most people will suffer in a lifetime when it comes to medical intervention.

Furthermore, because MLD is an auto recessive genetic disease, adult siblings of the parents of the affected children have the opportunity to be tested to determine if they are carriers as well. Two of Sonya's younger sisters are still having children, and after the boys' diagnosis, they were tested to see if they were at risk for having children with MLD.

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In conclusion, I believe that providing NBS for aggressive diseases, whether they are curable or not, is critical. Technological advances have provided us with the tools to detect diseases early on and as I've shown, the benefits of an early diagnosis to the child and his or her family are significant. Through NBS, you have the power to ensure that families and children do not have to needlessly suffer from misdiagnosis, frustration, and limited treatment options. I encourage you to consider this as you make your NBS decisions.

Sincerely,

Bob Evanosky
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