

# Stephen

THIS IS A PERSONAL STORY SHARED BY A FAMILY WHOSE CHILD HAS  
ISOVALERIC ACIDEMIA (IVA)

My name is Tom Monaco. My wife Jana and I live in Lake Ridge, Virginia (Northern) and are proud parents of 3 boys. Nicholas (10), Alex (7), and Stephen (3 1/2). Our three boys were normal, happy, and healthy until tragedy struck our youngest boy on May 30th.

On May 29th, Stephen was having a bout with a little virus. Some vomiting, but nothing more. Seemed normal, nothing to get too alarmed over. He finally went to sleep that night for good around 2:00 AM. When my wife went to wake him the next morning around 9 AM, she found a completely non-responsive boy. We immediately called our pediatrician who recommended an immediate trip to the hospital.



When we got to the hospital, the Doctor's there, after getting initial readings on his glucose and oxygen levels became very concerned. We explained to them that something similar to this happened 2 years ago, but then after a night of IV fluids for his dehydration, he came out of it and was okay. They said it was just roto-virus back then. However, this time, they said Stephen should be transferred to Inova Fairfax hospital in Fairfax, VA where they have a state-of-the-art Pediatric ICU because they felt Stephen needed that type of monitoring. This all started to become very scary for Jana and me.

When we got to Fairfax, they too started to suspect something more was wrong with Stephen and began getting the appropriate people involved. A geneticist was brought in to start asking family questions, blood tests were ordered, and so on. My wife and I all the while were just waiting for the IV fluids to "kick-in" and Stephen would wake up and start to get better as before. Well, the next few days turned out to be the worst days of our lives. The next day, when they were preparing Stephen to go for a brain MRI, he went into the first of several seizures. When he came back from the MRI, Stephen "crashed" on us and the Doctors said he was nearly declared "brain-dead" on the spot. But they got him stable by intubating him and helping him to breathe. The first MRI came back with a result of swelling on the brain stem. That same day, the Doctor's finally found through the blood tests what Stephen's problem was. He was diagnosed with Isovaleric Acidemia. As I'm sure you know, this disorder deals with the protein, Leucine. The virus apparently burned up the protein his body did have stored, and created a very acidic child. This acid running through his blood triggered the seizures and subsequent damage to the brain. At the time, they said they would just try and keep him stable through the weekend and hope this medication (name eludes me now) would help to reduce



the swelling on the stem and see how he reacts. They warned us however that our boy would never be the same again.

A second MRI was performed on that Monday, June 4th which revealed the most devastating news to date. It showed severe damage to his Cortex, Brain Stem and Cerebellum. At the time, our Neurologist and Pediatrician gave us no hope of survival, or if he was to survive, it would have to be completely on life support systems. You can imagine the devastation this brought to my wife and I.

However, through the help of a final month pediatric resident, and I will mention his name here because he is truly a man placed in our lives through God, Dr. Dan Feldman, was having a hard time believing that Stephen's life was over. First of all, there are very few documented cases of Isovaleric Acidemia. They said 70 at the time since 1989, and even less than half of those cases would present themselves for the first time after the age of 1, making Stephen's case all the more rare. So, being "only" a resident, he could only advise us of things he felt we could and should do. He truly picked us up after the dust had settled, brushed us off and got us focused on being the advocates for our child. We went out and sought 2nd opinions from pediatric neurologists from Children's hospital of Washington, DC, we went and found other "experts" of acidemia's like those at Baylor University Children's hospital and at John Hopkins. But all in all, no one gave us much hope. We allowed time to pass to see if Stephen would show any change. The changes that came were very minor and were chalked up to pure "reflexes" according to the neurologists.

At the two-week mark, my wife and I were at our wits end. We traveled down many roads for opinions with no reason to hope, we prayed together and sought spiritual advice from our priest, and if we chose to shut down his support system, that would have been okay because we had done all that we could. Then the next day changed things forever. A Psychiatrist named Dr. Shin, who was out of the country for the past 2 weeks came to visit Stephen. Prior to this, one of his partners was in to see him, but was obviously very junior to Dr. Shin and didn't really commit to any level of rehab that would benefit Stephen. Dr. Shin looked at Stephen's records over the previous two weeks, and came in to see me. He said based on his experiences with brain recovery, especially involving children, that Stephen had shown enough signs of change, that he could indeed benefit from rehabilitation and stimulation. He explained how he believed the brain can recover to some degree from these injuries.

From that day forward, Stephen showed significant signs of improvement to where two weeks later he was removed from the ventilator and was breathing comfortably on his own. He is fed through a G-Tube, now has finished a stay at Kluge Children's Rehabilitation Center in Charlottesville, VA. He is no longer in a coma. He has a definitive wake and sleep cycle. Both of his eyes are open when he is awake. He is starting to react to some of the therapies. For instance when his hands are placed into stimulating things like a box of marbles or sand, he reacts very negatively pulling his hands out and crying. Very positive signs.

We started him on a relatively new drug named Mirapex which according to our Pediatric Neurological Psychiatrist, Dr. Peter Patrick, could generate some of the undamaged portions of the brain to take over some functions of the damaged portions. We started with a drug called Amantadine, but he reacted very negatively to that so we stopped and started with Mirapex. At Kluge, they have had several children over the past 2 years placed on these medications (none with brain injuries due to disorders however), but 5 of those 7 have showed some level of progress because of it. One example they use is a 17-year-old boy who was a near drowning victim who was left in a complete vegetative state with a tracheotomy and G-Tube needed for survival. After a year of taking this medication and rehab, regained some walking with assistance and some level of speech. So we continue to hope and prayer for positive things for our Stephen, but we are already light years from where we once were 2 months ago and we feel blessed by that. We have prepared ourselves the best we can to take and care for Stephen in the state he is today, but will never give up hope of better things for him.

I share this story with you because my wife and I have had the opportunity to do some research and we have found this web site and it seems to be just what we need. We obviously have a long way to go, but I know we would like to get involved with screening awareness and local support groups if they exist. Virginia, like many other states is definitely way behind the times and does not screen for these types of disorders.

So my first question is, is there a support group/organization in the Northern Virginia area on Organic Acidemias? Being this close to Washington, how can we help petition the passage of laws for these screenings and what has been done to date? Any and all information about how to get involved would be appreciated and on support groups with folks going through similar difficulties.

My wife and I continue to go through the grieving process. I am the "baby" in my own family, so I had a very special bond with my baby and feel very cheated. Through faith, I am learning to deal with this and actually am beginning to feel blessed that we may be able to help fight the ignorance that is obviously shown in this country on these disorders and want to help bring the awareness to the level it needs to be. If Stephen is a strong example of where the proper screening would prevent others from being where he is now, then I think we would like to use that example to fight this fight.

Finally, through all this, we just found out that my wife is pregnant with our 4th child. Although not planned, we find this to be a sign and blessing. We are very excited about it. There can be no other answer for the bizarre twist our lives continue to take.

I have found some of the stories on the web site both sad and inspirational and want to be a part of it. Thanks for taking the time to read this story.

Sincerely,  
Tom Monaco

PS: The newest addition to the Monaco Family is Miss Caroline (IVA). She is doing very well today, due to prenatal diagnosis.