The Consumer Advocate's Voice

Mountain States Genetics Regional Collaborative

Gina Pola-Money: Rare Genetic Syndrome Diagnosis

I was a very young mom when I delivered my first baby by an emergency C-Section at the end of June many years ago. Tyson was born weighing only 4lbs.11 oz. but beyond being tiny, he seemed perfect in every way. After getting home his first 6 months proved to be challenging, he was constantly sick and we would be at the pediatricians office or in the ER a few times a month with respiratory distress. Even though I was new to all of the gifts and challenges of being a first time mom, I knew in my gut that something was just not right. Every time he became sick, his ability to kick his legs or roll over became harder and harder.

At about 7 months of age he was taken to the Children's hospital by ambulance from the pediatrician's office and this time it was different, he could not breath at all, they intubated him and unfortunately that was the last time I was able to see him with out a machine breathing for him. It took many months in the PICU before it was confirmed that he wasn't contagious and was moved out of the isolation room but there were still no answers as to what was going on. All of the tests and procedures would come back inconclusive and as young parents we were left with realizing that we may never know a name of the monster of a disease that had taken the ability of my baby not to roll over let alone breathe. I really needed something to blame! Tyson was eventually sent home fully ventilator dependent. We all thought that we were taking him home to give him all the love possible because he probably would not survive very long due to the unknown condition.

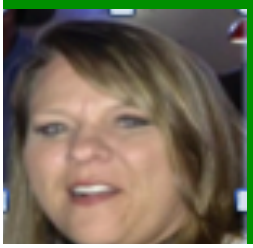
He did survive and thrived even though he had limitations due to the ventilator and the progressive muscle weakness. We continued with our family after four years of limited information and the unknown. Our family settled into what was "our normal" life with Tyson. I was blessed with two more boys and a girl. As our life progressed we did eventually get a diagnosis for Tyson, it was Neuroaxonal Dystrophy, which is very rare. I finally had something to blame but what I didn't understand at the time was it was a recessive genetic condition and that there was a 25% possibility of having another child with the same diagnosis. I felt somewhat safe though because the other three children were not sick all the time like Tyson once was but that feeling soon left and that same fear of loss came again and again. My husband passed away suddenly from unknown flu like symptoms.

After settling into again what would be "our normal" I noticed that my third son Bryton was not keeping up with the developmental milestones. With some testing and guidance from our Neurologist it was determined that Bryton also had the same monster of a disease that Tyson was living with, it just presented in a totally different manner. The fears of losing one or both of my children and wanting to protect them from any pain and suffering was overwhelming.

Bryton passed away just shy of his 5th birthday, we chose not to do any invasive measures with him to honor his wishes, he was able to communicate how painful the disease was and how much he did not want to be put on "tubes" like his older brother. Tyson passed away at the age of 17 after many years of giving us much more than we could ever have given him. Both boys are and will continue to be my heroes and the reason I will continue to advocate for a system of care that is easy to navigate regardless of the diagnosis.

Families need as much information as quickly as possible to make informed decisions that not only impact the lives of our beautiful children living with genetic conditions but for those that love them unconditionally and are living the day in the life.

The road to services and financing was and still is very fragmented but we got through it because we had professionals that were a part of "our" team and they supported the fact that we were the experts when it came to the decisions about hopes, dreams and family-centered outcomes.



Gina Pola-Money decided to put forth her time and energy in helping to advocate for healthcare financing and adequate family resources over the last 25+ years. She has dedicated much of her work to Medical Home projects and community based systems of care and resources. She has worked with the Bureau of Children with Special Health Care Needs for the last 13 years and also have had the opportunity to be the Director of the Utah Family Voices, Family-to-Family Health Information Center since the beginning. She has been honored to be a Consumer and Medical Home Committee member of the Mountain States Genetics Collaborative over the last