Public Comment to the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children

Jana Monaco

Organic Acidemia Association and the National Coalition for PKU & Allied Disorders

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Good afternoon ladies and gentlemen. It is an honor to be here today on behalf of the Organic Acidemia Association and the National Coalition for PKU & Allied Disorders. Most importantly though, I am here for my 6 year-old son Stephen, the youngest of three boys, and to tell you about the harsh reality of undetected inborn errors of metabolism.

You see, ironically, 3 years ago today, I was sitting in Stephen's ICU room alongside my husband, trying to determine whether or not to discontinue life support. Ten days earlier, Stephen had contracted a typical stomach virus, like most children do, and I treated it as such. However, I found him the next morning in a state that no mother should ever have to endure. He was lying in his crib, breathing incredibly fast with his eyes half-way opened with a distant gaze. He was completely unresponsive. Stephen was transported to our local hospital and then to Inova Fairfax Hospital's PICU. Stephen's tests revealed severe acidosis leading them to a diagnosis of a metabolic disorder. The initial test eliminated certain disorders, but others had to be sent out. Twenty-four hours later, Stephen was diagnosed with Isovaleric Acidemia, in which his body's enzyme to break down the amino acid Leucine, either doesn't exist or does not function properly. Leucine produces Isovaleric Acid and when those levels build up in the body, they can become toxic. Unfortunately, Stephen's diagnosis came too late. By the next day, he was slipping into a coma. While preparing for an MRI Stephen went into a grand mal seizure, followed by another one in the MRI. Within minutes of returning to his room, Stephen crashed before our eyes. After a great deal of intervention, Stephen was clinging to life on a respirator, with a central line and chest tube placed. It is a sight that remains etched in our minds today. The initial MRI indicated swelling around the brain stem. The weekend only brought reflexes from Stephen and another MRI that Monday, revealed extensive brain damage throughout Stephen's brain. That's when we were faced with the decision of discontinuing life support. As you can imagine, we were devastated with the news of losing our son. We asked "how could such a happy, healthy, energetic, child become so close to death in such a short time? While we were trying to come to terms with Stephen's condition and prognosis, we discovered that he was a walking time bomb waiting to ignite and that this whole situation could have been avoided had he benefited from Comprehensive Newborn Screening at birth. We also linked a similar episode at 18 months to the disorder, but the doctors failed to recognize the signs and symptoms of the disorder. The physicians and hospital had acted within the standards of care for a small community hospital. Hindsight is brutal. Looking back, the signs were all there…strange odor, picky eating, and slow weight gain. "You had no way of knowing ," is what we were told. We gave Stephen a little more time and he started to show signs of progress. After 3 and a half weeks, Stephen received a gastrostomy tube and was removed from the respirator. A week later, he was transferred to the Kluge Children's Rehab Center in Charlottesville, VA. There we spent 6 long weeks where he endured daily therapy and care to get his body well again.

Since then, Stephen has made progress that was not to be expected. However, he is far from the little boy that he once was. He requires total care. He continues to be fed via gastrostomy tube, and he can not walk, talk, sit up, nor hold his head up without support. He also is legally blind. Stephen takes four anticonvulsant medications per day, yet still has 3 to 4 seizures a day. Due to his neurological state, hiccups usually result in a hospital stay, because his button causes GI bleeding. He recently had surgery called an orchiopexy, which is a surgical procedure to bring his testicles down. They had retracted this past year due to spasticity. Our days are filled with therapies and numerous doctors' appointments. I spend many phone hours trying to settle insurance disputes. His medical costs have exceeded the million-dollar mark and continue to climb. Stephen is now under the school system, where he has an IEP. This too is a new battle, to ensure proper care and services for Stephen. He has already outgrown his first wheel-chair, and we are awaiting another that will cost approximately $5000. We would have much

rather paid the $25 to $40 for Comprehensive Screening had we only known that it existed. Gone are Stephen's opportunities for a normal life, because our government and health system continue to debate the cost effectiveness of Universal Newborn Screening. The Children's Health Act of 2000 had promised to help fund States expanded newborn screening, but has yet to follow through with the money. States are left to their own means and only 18 have decided that children's lives are worth the effort and cost. Stephen's fate was already determined because he was born in Virginia, where they only screen for eight disorders. Had he been born in our neighboring North Carolina, where the list includes 36 disorders, Stephen would be in a normal kindergarten class instead of occupying a special education slot. It is a travesty that Stephen is a statistic at the hands of beurocracy and lack of knowledge within the medical community. While the debate continues more babies are going to die and more children are going to share Stephen's fate, yet the equipment and knowledge to avoid this exists. The life of my Stephen and the thousands like him born each year should not be so devalued in a society where our constitutional rights are supposed to promise us equality. The incidences of these disorders are not as high as other diseases like cancer and diabetes, but they can be just as debilitating and deadly. Most important of all, the treatment for most of these disorders already exists.

A testimony to the significance of early detection of these disorders is our 20-month-old daughter, Caroline. With the knowledge we gained with Stephen, Caroline was diagnosed with the same disorder with prenatal testing. Early diagnosis enabled doctors to establish a protocol of care prior to her birth. With a restricted protein diet and medication, Caroline is doing well and developing normal. She is a typical, happy, healthy, toddler thanks to early detection. Unlike Stephen, she will have a normal childhood and dreams. Although Stephen has suffered severe brain damage, and dreams have been lost, we know that his life has a purpose and we will see to it that it is fulfilled. Thank you for your time.

Jana Monaco
3175 Ironhorse Dr.
Woodbridge, VA 22192