

DYSAUTONOMIA INTERNATIONAL



AWARENESS



ADVOCACY



ADVANCEMENT



**21st Century Cures Roundtable
Tuesday, August 19, 2014 Lutz, Florida**

Testimony Submitted by Ashleigh & Beth Pike, Dysautonomia Patient Advocates

Good afternoon Congressman Bilirakis. Thank you for having us. My name is Ashleigh Pike and prior to August 2, 2013 I was a healthy 25-year-old about to begin my 5th year teaching elementary school. I loved to dance and was a regular at the gym. While on summer break, I traveled to Washington D.C. for a Beyoncé concert with a girlfriend and came home in a wheelchair. I had a grand mal seizure and spent 9 days in the ICU resulting in the inability to stand up without fainting.

Over the next year, my journey of body failure and unanswered questions took away my career, my brand new home I had recently purchased as well as my mom's ability to work as she had to become my full-time caregiver on our "adventure" across the country to find answers. We were medi-flighted to Vanderbilt hospital in Tennessee after spending a month in hospital in Countryside. After being sent home and told by doctors that they had done all they could do, we spent months alone trying to figure out what to do without much help from any local doctors.

My parent's persistent advocacy has helped in determining a possible diagnosis. Others wait years due to the rarity of different Dysautonomias. The irony of this debilitating disease is that I look fine on the outside, however internally, my body is attacking itself. My quality of life has been greatly compromised. I have spent my 26th year of life almost entirely laying down, seated or standing for a consecutive period of about 20 seconds or less at a time.

If I did not have the support of family, church, coworkers and friends, this experience would be complete misery. I am blessed. This life lesson has been anything but easy. However, I am aware that there is an opportunity for my suffering to possibly help bring answers and/or awareness to guide or assist others with rare diseases. That is why we are here today and we are so grateful for your listening ears and open hearts.

Hello, I am Beth Pike, Ashleigh's mom, and as you just heard we are dealing with a rare disease with limited treatment options and no cure. I'd also like to thank Representative Bilirakis for giving us the opportunity to speak here today and the Energy and Commerce Committee for introducing the 21st Century Cures Initiative.

We are here today representing all of the patients who have chronic, complex, and disabling forms of Dysautonomia, which is likely the most often overlooked, misdiagnosed condition our time. Dysautonomia is a failure at some level of the autonomic nervous system. Think of all the amazing things your body does without you thinking about it, or what happens automatically like breathing, heart rate, digestion, sweating, and blood pressure. Dysautonomias can stop these automatic and essential functions from happening.

Neurocardiogenic Syncope is a common symptom of many Dysautonomias which causes fainting due to severe drops in blood pressure. These drops are sudden; some patients fall and suffer from broken bones and even brain injuries, others cannot even get out of bed. There is a huge spectrum of degrees and complications from Dysautonomias. Some are more common but some, like Ashleigh's are extremely rare. Specialists have yet to even agree on a specific diagnosis for Ashleigh because of her unique symptoms. The world's expert in autoimmune sensory and autonomic ganglionopathy Dysautonomias told us he has seen maybe 4-5 patients who present like Ashleigh.

Multiple System Atrophy or MSA is a fatal form of Dysautonomia and people generally die two to five years after being diagnosed. There are many more forms, which complicates diagnosis and treatments, but all of these forms can leave a patient with reduced ability to work, attend school, or participate in social activities. It is estimated that more than 3.5 million people in the U.S. have a chronic form of Dysautonomia that remains undiagnosed today.

Difficulties patients face are numerous. Getting diagnosed is the first battle. Most patients take an average of 6 years to receive a proper diagnosis because of a lack of awareness and understanding among medical providers. We spent 3 weeks at Mease Countryside hospital and another 4 weeks at Vanderbilt Medical Research Center and still did not receive a definitive diagnosis. Two of those weeks, we were waiting without any treatment or advice from a doctor because of insurance issues and the difficulties to get her transferred to a different hospital. We lost that window of opportunity that possibly could have made a difference had the proper treatment been given right away.

We were fortunate that we had family support that allowed us to afford to travel to places like Vanderbilt and Dallas for Ashleigh's care. The majority of doctors here in the Tampa Bay Area have told us they are unable to help due to the complexity of her case. We have heard stories of another family who were told there is nothing wrong with their daughter. The doctors prescribed Xanax and told her to see a counselor. She also had to travel to the Cleveland clinic, Vanderbilt and Charleston for a diagnosis.

Once a patient is diagnosed then another battle has to be fought. Finding a physician who is willing to work with out of town specialists is often difficult. In Dysautonomia like many other rare diseases, the symptoms affect several organ systems. But our health care system is set up to address specific regional areas of the body. So when you go to the cardiologist he looks at the heart and circulatory system without considering the neurological, gastrointestinal, or pulmonary effects. In order to better treat Dysautonomia patients we need specialty physicians who look at chronic and complex issues involving multiple organ systems. David Goldstein MD, PhD at the NIH has proposed this as "Scientific Integrative Medicine."

Once we find a physician who is willing to work and learn with us, gaining access to affordable medications can be extremely difficult. Many times insurance companies won't cover them because most of the medicines are used "off label." Meaning there are not tested or approved for our disease, but they are our only option.

We need Congress's help to ensure patients have access to affordable treatments!

- 1) The Orphan Drug Act was passed which provides financial incentives to encourage development of new treatments for rare diseases, however we need more incentives because so many patients are still without treatment.

For example, Droxidopa which was approved by the FDA this past February is still not available for Dysautonomias patients. This drug has been used in Japan since 1989! We are happy the FDA approved

this very important drug used for increasing blood pressure, but are concerned because Dysautomias patients have to use the drug "off label." This means the insurance company will not reimburse for this drug. Because Dysautomias are rare there is no financial incentive for a drug company to study the drug in our disease or to repurpose the drug for use in our patients.

A solution would be a simple economic incentive for industry to repurpose major-market patented drugs for rare diseases or "Rare-Purpose" drugs. An incentive could be modeled after the Best Pharmaceuticals for Children Act (BPCA) to give companies an additional 6 months of market exclusivity to life of their drug patent which has been proven to be very valuable to companies. An "Orphan Product Exclusivity Extension" could make hundreds of safe and effective drugs available to patients like Ashleigh in the next few years. It is estimated that 120 drugs go off patent every year, so every year we lose these opportunities to rare-purpose these drugs.

We are asking Congress to include a new market exclusivity extension for rare-purposing drugs in the 21st Century Cures Legislation.

- 2) Another concern is the FDA has threatened to pull Midodrine off the market because the drug company that obtained the original approvals never finished their post-marketing efficacy studies. The patent has expired so there is little economic interest for the company to fight to keep the drug on the market. Almost 100,000 Americans, including Ashleigh, use Midodrine everyday just to be able to get out of bed and stand up. Ashleigh is one of those Americans. There are no other alternative drugs that can do what Midodrine does at this time.

We need the Congress's help to make sure the FDA does not pull this drug from the market. The FDA should allow for flexibility and should be willing to look at studies done outside of an FDA submission.

- 3) The FDA drug review divisions that approve rare disease drugs need to have experts on the particular rare disease serve as ad hoc members of the divisions. Having a bunch of doctors who have never diagnosed and treated a particular disease deciding what drug should or shouldn't be approved makes no sense! There should also be at least one patient advocate expert for the rare disease to serve as an ad hoc member as well.
- 4) We need the Congresses help to restore funding to the Autonomic Disorders Consortium, previously funded by the NIH Rare Diseases Clinical Research Network. The Autonomic Disorders Consortium is hosted by Vanderbilt and it is the best collaborative autonomic disorders research program in the world. Just last week the NIH decided not to renew funding for this program, which gives hope to all Americans living with rare forms of Dysautonomia. The Consortium has been funded with 5 year grants of about \$1.2M per year for the past 10 years, and it supports the essential collaborations between Mayo Clinic, Vanderbilt, Harvard, NYU, NIH and other leading Dysautonomia research centers needed to conduct research on rare autonomic disorders. No one research center sees enough of these rare disease patients to do research alone, so they have to work together. Without the Autonomic Disorders Consortium to support collaborative research on rare forms of Dysautonomia, this research will come to a standstill, leaving patients with little hope for better treatments in the future.
- 5) Finally, we need your help in supporting The Dysautonomia Project that is launching here in the Tampa Bay Area on October 9th. We hope you will help by attending and encouraging community leaders in

their districts to participate in local fundraising events that champion physician education and community awareness. The aim is to speed the time to diagnosis and train frontline physicians locally and across the country to properly assess and treat Dysautonomia.

We would like to thank everyone who came out today to help support us and we would especially like to thank Representative Bilirakis and the Energy & Commerce Committee for supporting patients with rare diseases and making a difference.