

February 20, 2015

Re: HB 6580, AN ACT ESTABLISHING AN ADVISORY COUNCIL ON RARE DISEASES

Testimony for Public Hearing
State of Connecticut Public Health Committee

Chairman Ritter, Chairman Gerratana, Members of the Public Health Committee:

I am a physician who has specialized in the area of metabolic disease for 29 years. I served as Director of the Inborn Errors of Metabolism Service at Boston Children's Hospital for until the year 2000. I then became Chief of the Metabolism Service at the Floating Hospital for Children at Tufts Medical Center in Boston, a position I held for 14 years. My work included the diagnosis and treatment of inherited metabolic disorders. Currently, I am Medical Director of the Genetic Metabolic Center for Education.

More than 25 million Americans and their families are affected in the US by one of over 7000 rare diseases. Patients affected by rare diseases can present with a variety of symptoms, ranging on a spectrum from mild to severe. Rare diseases often cause chronic, progressive illness leading to disability and sometimes an untimely death. The exact cause for many rare diseases is unknown. A majority may be caused by some genetic alteration; environmental factors may also cause disease or trigger the presentation of an inherited disorder. Rare disease affects every age group, culture, and socioeconomic status.

Diagnosis and treatment for a patient with a rare disease can be complex because medical students and physicians are not well trained in this area. As a result, symptoms may not be recognized and patients are frequently undiagnosed or misdiagnosed. This leads to excessive and repetitive testing which can be very expensive. Although definitive testing such as genetic testing may be costly, it may save money in the long run by shortening the duration of testing and reducing the number of tests to be done. The lack of a diagnosis can be tragic when treatment is available; without a diagnosis, that treatment is inaccessible. Furthermore, without a diagnosis, prognostic counseling cannot be provided which gives the patient and family an idea about future expectations and information about potentially preventable complications yet to come. Furthermore, the lack of a diagnosis means that families cannot be provided genetic counseling, i.e., information about the risk of having another affected child;

the reproductive risk remains unknown. On the other hand, available treatments can be expensive and medical insurance companies often withhold approval so that patients are unable to afford the chance for a better quality of life.

It may be challenging to locate a specialist who knows how to diagnose and treat a rare disease since qualified physicians are few and scattered across the country. Insurance companies do not always recognize the need for specialized medical care and often deny patients the access to physicians with expertise in their symptoms or disease.

Compounding these issues of lack of awareness and obstacles to care are the limits in funding to develop useful diagnostic tests and effective therapies. Inadequate research impedes the opportunity to give this patient community a better quality of life. Funding opportunities for research is a paramount need.

I have learned a great deal from my patients and their families. They have taught me to see past the symptoms of their diseases. They have shared with me their frustrations and agonies, the challenges they face on a daily basis. I listen patiently and offer a compassionate tone but this community deserves more than this. They deserve to know what ails them using up to date testing. They deserve access to effective therapies. They deserve the knowledge that can help them understand what the future holds and what their reproductive risks are. In short, they deserve the same quality of life that we all come to expect.

With the proposed legislation, CT will have the potential to recognize problem areas in the care provided to this patient community, develop solutions, and make changes that can benefit their lives. I urge you to support this proposal to establish an Advisory Council for Rare Diseases.

Thank you for your time and consideration.



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