

Testimony for Public Hearing
Public Health Committee
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HB6580, AN ACT ESTABLISHING AN ADVISORY COUNCIL ON RARE DISEASES.

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

My name is Julie Gortze. I am an RN and I have cared for many patients living with disease. I have seen these patients in their homes, schools, rehabs, and in acute care settings. I am a patient advocate for people affected by rare diseases. I am very active in the community with regard to those touched by rare diseases as I provide support, offer knowledge of resources available, and become involved in areas to make better outcomes for quality of life for the rare disease patient. I have contact with various rare disease organizations. I collaborate with physicians who focus in diagnosis, treatment and research of rare disease.

I am here today to ask for your full support of HB6580, An Act Establishing an Advisory Council on Rare Diseases. I envision such a council, consisting of members who represent several disciplines involved with rare disease, including, but not limited to, physicians, social workers and school officials, as well as adult patients, parents, and rare disease organizations, gathering to compile and evaluate the issues in rare disease; then moving forward to rectify the difficulties which keep those affected from acquiring a better quality of life.

Although there are over 7000 rare diseases, with numerous variations of symptoms and levels of severity, the rare disease patient is plagued by many common adversities. The facets of living with a rare disease are far too complex to satisfactorily explain in the short time for this testimony. Let me highlight a few specific cases where I have witnessed conflict due to barriers for someone living with rare disease:

- A family having to get up 2 1/2 hours early each morning to prepare for the young son's day, administering medications, changing feeding tube connections, and more - to then put him on a bus that included an hour and a half ride EACH way to his specialized school.
- A mother who called in to a support hotline to ask for advice on which of her two daughters, both of whom were diagnosed with the same rare disease and prescribed the same medications that insurance would not cover, should receive the meds that month as she just didn't have the income to cover both.
- A middle aged woman who became stranded far from home as she had taken a commuter train and found the walkway to the train upon return did not accommodate for her energy disorder.

- A woman who was accused of taking recreational drugs as cause for her acute symptoms by ER staff because they did not understand the complexities of her disease. This prevented appropriate medical care in a timely manner.
- A young adult who is suspected of having a complex rare disease who is unable to see a specialist in that disease because there are too few who treat that particular disease. This patient also cannot obtain definitive diagnosis because the only chance for that would be \$16,000 DNA testing of which his medical insurance refuses to help pay for. This young man has little quality of life as he is too ill to get out of bed most days.
- A middle-aged man who has a confirmed diagnosis of a rare disease, poor quality of life due to symptoms which make him a poor work candidate yet disability denies him because they don't understand the repercussions of his disease.
- Physicians, nurses, social workers, and other medical staff stand by helplessly as their patients lose body system functions and then succumb to their diseases of which there is no adequate treatment, knowing full well there is none or insufficient research being done to find a cure for their disease, due to lack of funding.

These are just a few samples of what I have witnessed. As you can see, there are a great many aspects in rare disease which need addressing. These scenarios barely scratch the surface. I ask that you give this bill, and all its potential, serious consideration. I feel a Rare Disease Advisory Council would provide an access to recognize, investigate, and then implement changes accordingly which could benefit the whole rare disease community.

Thank you for your time today.

Sincerely,

Julie Gortze, RN

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Bullet points of submitted bill language from MA rare disease bill of which I would like to see for goals in CT as well:

- Coordinate statewide efforts for benefit of those affected by rare disease, DPH guided
- Consisting of several reps of pertinent medical specialties, rare disease groups and organizations

- Examine incidence and cause of rare disease
- Develop profile of social and economic burden of rare disease
- Develop methods to publicize the profile of social and economic burden of rare disease
- Consider reports from those affected by rare disease, including individuals, health care providers, and public and private organizations to learn possibilities for improvement for rare disease care.
- Identify research-based strategies to prevent and control risks of co-morbidity for rare disease
- Determine the impact that delayed or inappropriate treatment of rare disease has on patient's lives and economic burden on both patients and the Commonwealth.
- Study the economic burden in regard to quality of care, insurance reimbursement, and rehabilitation.
- Develop ways to ensure that the public and health care providers are informed in recognizing and treating rare disease
- Evaluate current rare disease treatment for purpose to raise potential of increasing rare disease survival rates and improving quality of life
- Research and determine most appropriate way to collect rare disease data
- Identify best practices for rare disease care by other states and at national level
- Propose structure for developing a patient support network
- Identify sources of funding to improve rare disease care
- Develop a registry of rare disease diagnosed in the commonwealth to aid in determining environmental or genetic factors
- Develop and maintain a comprehensive rare disease plan for the commonwealth, including providing materials to be utilized

