

Testimony for Public Hearing

Public Health Committee

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As the father of 3 children with rare diseases, I am entirely supportive of the establishment of a state council on rare diseases. The diagnosis and treatment of rare diseases is overwhelmingly complicated and resources to assist those whose lives are impacted by rare diseases are extremely deficient. It is my belief that the lack of a central resource that could help families and doctors navigate the complex process of diagnosis and subsequent disease management results in poorer health outcomes, decreased quality of life for those suffering from rare diseases and increased cost to payors, including the state.

One of my children has a condition, that while rare, is included in the newborn screening in CT and has been definitively diagnosed. We were notified of the results of the newborn screen just as our daughter was slipping into a metabolic crisis and the screening may very well have saved her life.

Our other two children also suffer from different rare diseases that have proven more difficult to diagnose. In our case, we, the parents, were the first to suspect that something was not quite right. It took determined advocacy by the parents to progress down a series of tests including fasting studies, multiple day continuous blood sugar monitoring, liver biopsies and muscle biopsies to confirm that there is a very real underlying physical cause for the developmental and physical issues that were the cause of our concern. Along the way we were referred to experts in various fields including geneticists, endocrinologists, neurologists, nutritionists and metabolic specialists. This journey took several years and would have taken longer without the determination of parents who continued to push for a diagnosis. In the absence of a diagnosis and an appropriate treatment plan, our children's development continued to be negatively impacted by their disorder. If a centralized resource had been able to help identify and proactively coordinate the process of diagnosis and development of a treatment plan, the long term impact on our children's lives may have been mitigated and the costs, associated not only with the diagnosis process but with ongoing medical care that will be required throughout their lives may have been significantly reduced.

The body of knowledge within the medical community is expanding at an unbelievable pace. Based on our experience, however, that knowledge resides in pockets of expertise that are not well coordinated and as a result it is not applied in a timely or efficient manner. This fragmentation is confusing and exhausting to navigate, has a negative impact on the health outcome of patients and is almost certainly unnecessarily expensive both in the short and long term. A rare disease council that could serve as a resource to both families and the medical community could be amazingly beneficial and represent a vast improvement over the current system.