

State of Connecticut
GENERAL ASSEMBLY



PUBLIC HEALTH COMMITTEE
LEGISLATIVE OFFICE BUILDING, ROOM 3000
HARTFORD, CT 06106-1591
Telephone (860) 240-0560 Fax (860) 240-5306

Task Force to Study Rare Diseases

Meeting Summary

February 24, 2017

Dr. Gulati and Dr. Lutz convened the meeting at 1:45 PM with introductions of the Task Force members.

Members Present: Dr. Gulati, Dr. Lutz, Lesley Bennett, Nancy Dupont, John Morthanos, Lynne Sherman, Dr. Joe Tucker and Paul Pescatello

Dr. Gulati briefly covered the Orphan Drug Act of 1983 and the Rare Disease Act of 2002 which established the Office of Rare Diseases. These Acts promoted a national research agenda, coordinated research, and provided educational activities relating to rare diseases.

Dr. Gulati stated that there are 7,000 rare diseases in the United States affecting less than 200,000 people. 25-30 million people in the US or about 10% of the population suffer from a rare disease. 50% of those with rare diseases are children. Most rare diseases are genetic and lack cures or treatment.

Dr. Gulati provided an overview of the legislative charge of the Task Force which is to; (1) examine research, diagnoses, treatment and education relating to rare diseases, and (2) make recommendations for the establishment of a permanent group of experts to advise the Department of Public Health on rare diseases.

Dr. Gulati asked Task force members to briefly discuss their priorities and goals for the Task Force.

Lesley Bennett, a patient advocate, stated her focus was political and wants the Task Force to become a permanent one appointed to the Public Health Committee. She stated that the Task Force focus should be helping Rare Disease patients with legislation for out of pocket costs related to specialty tier drugs. She also wants to focus on increased newborn screening that cannot only save lives but save all involved parties money in the long term.

Nancy Dupont, a Registered Nurse, stated that she would like to see the group focus on community outreach efforts to engage patients and the general public about rare diseases. She also made herself available to fulfill whatever role the Task Force asks of her.

John Morthanos, a rare disease survivor, stated that the Task Force should focus on finding a way to have a better informed medical community so they can be of greater help to the public.

Lynne Sherman, a pediatric nurse, wants to focus on providing patients with the right care at the right time with the right services, as well as pulling together the many different parties in the rare disease community and getting everyone on the same page and working together.

Dr. Joe Tucker, a Medical Geneticist, wants the group to increase access to services, foods and formulas for newborns with rare diseases that are often times very expensive and out of reach for the average patient.

Paul Pescatello, CBIA Bioscience Growth Council, wants to see the group focus on increasing funding for research and development to bring new drugs and treatments to the market. There is currently no cure for 95% of rare diseases and the only way to lower that percentage is to fund Research and Development.

Additional Discussion

Lesley Bennett stated that Xsome sequencing which can detect up to 30% of rare diseases is considered unproductive and expensive in the eyes of health insurers in the state and it is why they are hesitant to approve Xsome testing. She added that they prefer specific genetic testing.

Dr. Tucker stated that currently DSS offers various types of genetic testing through their Medicare program, although the application for such testing is burdensome and extensive.

Dr. Gulati asked the Task Force if local hospitals and providers have all the resources required to do genetic testing and screening of newborn babies before contacting a higher specialty care doctor.

Lesley Bennett stated that the answer really depends on the individual provider. Across the board many doctors are not up to date on the latest in genetic development and there is certainly a learning curve to be overcome. That is why it is so important that there be greater education of rare diseases in medical school.

Dr. Gulati asked about early referrals for individuals with rare diseases.

Nancy Dupont stated that there is certainly a lack of ongoing communication with information between patients and healthcare providers.

Dr. Tucker stated many places lack the resources and information to effectively diagnose and inform the patients. Dr. Tucker supports educating providers about Newborn screening.

Lesley Bennett stated that the State of Connecticut needs to look at using telemedicine more and that we currently lag behind many other states. She wants to improve care given to rare disease patients that will save patients, provider's insurers and the state money in the long run. She stated that she would like a member of the Public Health Committee and the Department of Public Health to be a part of the Task Force to provide knowledge and data.

Paul Pescatello expressed support for hearing the input of the insurance community and getting their perspective.

Dr. Gulati announced the date of the next meeting will be March 31, 2017 with the time and location to be determined.

The meeting adjourned at 3:00 PM.