

State of Connecticut  
GENERAL ASSEMBLY



PUBLIC HEALTH COMMITTEE  
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**Task Force to Study Rare Diseases**

Meeting Summary

October 20, 2017

Dr. Gulati convened the meeting at 1:15 PM with introductions of the Task Force members and the presentations.

Members Present: Dr. Cathleen Lutz, Dr. Lynne Sherman, Dr. Mridu Gulati, Nancy Dupont, John Morthanos, Michele Spencer-Manzon, Lesley Bennett and Dr. Barbara Pober.

Others Present: Dr. Marie Egan, Dr. David Weinstein, John Hopper and Adrienne Hoffman, Kristen Angel.

Dr. Gulati informed the task force of upcoming meetings on Nov. 17th, Dec. 8th, and Jan 19<sup>th</sup>.

Dr. Egan presented on Cystic Fibrosis (CF).



presentation1020  
without movies.pptx

Lynne Sherman asked Dr. Egan what areas the Task Force can focus on to benefit her research of Cystic Fibrosis.

Dr. Egan stated that the area the Task Force should focus on is the transition period from pediatrics to adulthood. She added that it's very difficult to transition from a psychosocial, insurance, and job standpoint and that there is so much vulnerability during those phases. Dr. Egan stated that the community needs more individuals trained and available to handle these transition and that this type of care is time consuming and costly.

Dr. Gulati asked what role does community pediatricians play in this situation.

Dr. Egan stated that she has a good relationship with pediatricians and added that pediatricians are the first ones to meet with patients and their families before referring them to the center. She stated that she works closely with pediatric offices to coordinate care of patients and the dissemination of information related to patients. She added that this type of work is a team effort that requires a lot of effort from all involved parties.

Dr. Gulati asked about the interactions between Cystic Fibrosis Center and the Department of Education and how that information is sent to nurses and the school.

Dr. Egan stated that communication is tricky because it is medical information and that every family determines if they want the school to know about their child's health. She informed the group that the center figures out what works best for each family and their school district and that often times the Center will work with the school systems designated nurse to coordinate the care of the child.

Dr. Pober stated that the Cystic Fibrosis Foundation had a visionary approach for cloning the gene and said they would not be funded unless they agreed to work with other doctors and share their equipment and data to make progress on research.

Dr. Egan concurred with Dr. Pober and added that there is a national registry so that there is no small cohort and that everyone has to share their information, data and equipment with the rest of the community for the greater good.

Dr. Pober stated that from a geneticist perspective Cystic fibrosis is actually very common.

Dr. Egan added that there is more opportunity for wealthy families to drive the foundation and that the foundation started in the 1950s by families that wanted to work together and find solutions for their children's condition. She added that this decision has paid off in the long run as well.

Kristen Angel endorsed the importance of transition from childhood to adulthood and talked about her first hand with her cousin who deals with Cystic Fibrosis. She explained how her cousin was cared for by a pediatric doctor well into her 20s before transitioning to an adult team. She thanked Dr. Egan and her center for all the great work that they do.

Dr. Egan stated that families need adult providers from the very beginning because the expectation is that the patient will grow up and reach adulthood. She added that operating that way from the onset will make future transition much easier.

Dr. Kathleen Lutz presented on Mouse Repository and In Vivo Pharmacology.  
Insert Presentation here

***(Presentation File Needed)***

Lesley Bennett asked Dr. Lutz how many projects she is currently working on with researchers from Connecticut.

Dr. Lutz responded that she is working on approximately 50 projects on individual diseases that feature Connecticut researchers.

Dr. Pober asked if the decade it took to decode Spinal Muscular Atrophy (SMA) is due to the fact that it was a decade ago and we were not as technology advance or the more complex genetic architecture of the Survival of Motor Neuron (SMN) gene.

Dr. Lutz stated that she believed it was the opposite, and that she thinks the SMA/SMN was the target of NIH because it is a very drug-able disease. She added that due to so many approaches been taken toward finding a cure the medical community was able to learn what worked. Dr. Lutz stated that even if the therapy didn't work for SMA, the community was able to find out what it could do for other disorders and much was learned about gene therapy in general.

Dr. Pober asked Dr. Lutz to clarify if she thought it took longer due so many different approaches.

Dr. Lutz stated it took so long because the community simply did not know enough at the time and the technology was not there and it was all brand new territory.

Dr. Pober stated the X-some sequencing is amazing and revolutionary but the diagnostic rate is still under 50%. She added that if you look at the autism for example, depending on the pre-screening of the patient, the diagnostic pick up rate is no more than 10, 20, 30 percent maximum. She pointed out that there is still much room for improvement.

Dr. Lutz agreed that improvement is needed for whole gene sequencing going forward.

Dr. Gulati asked how the Jackson Laboratory partners with the academic intuitions in the state of Connecticut.

Dr. Lutz stated that the Jackson Laboratory is a non-profit organization and most of the interactions happen at the academic level. She added that they also the works with industries which is uncommon and that as reproducibility becomes an issue, industries are looking for a third party to validate their findings. Dr. Lutz stated that Jackson Laboratory has no skin in the game with the results of the testing, so we can be objective about their findings. She added that as a result of this, a level of trust is developing between her lab and the industries.

Dr. Gulati asked how the state collaborates with the Jackson Laboratory.

Dr. Lutz stated that it is important that all the research conducted by biotech and pharmaceuticals industries have their work supported and Jackson Laboratory help with keeping cost down and finding qualified staff to work at these places.

Dr. David Weinstein presented on the Glycogen Storage Program.



Weinstein Rare  
Disease Task Force.p

Lesley Bennett asked Dr. Weinstein to explain further about his clinic and the family atmosphere they have.

Dr. Weinstein explained that he is the most fortunate physician because he has the most dedicated team. He added that his entire team, with the exception of one staff member

moved with him from Florida to the new location in Connecticut. He added that the team is especially dedicated to the work they do because many of his staff have children or are actually patients with a rare disease themselves.

Dr. Gulati asked how patients find out about Dr. Weinstein.

Dr. Weinstein stated the internet is a very powerful tool and that organizations such as NORD are very important in disseminating information to patients. He stated that one problem is that he cannot take care of all the patients. He added very few American Doctors are interested in doing rare disease work because there is no money in it and hospitals do not want centers because it is not profitable, which results in many of the doctors being foreigners. He stressed the need for the U.S. to train more American doctors and to build infrastructure to support the rare disease community.

Dr. Pober asked what happens to the financial model when a treatment is not available.

Dr. Weinstein stated that when there is no treatment, families will support research out of desperation. He added that philanthropy should be easy for rare disease because sometimes there are only 1 or 2 foundations and in many cases none.

Dr. Pober asked if the state should be asked to co-fund the research or offer matching dollars to incentivize research.

Dr. Weinstein agreed with Dr. Pober and said other states have begun to do similar things in regards to research and matching private dollars. He added that Massachusetts does that with Boston Hospital and that it is a good way to foster research and investment.

Lesley Bennett stated that she heard about Dr. Weinstein's program from the Big Wheel Derby event in West Hartford, which sought to raise funds to send children and their families to see Dr. Weinstein in Florida before his relocation to Connecticut.

Dr. Weinstein stated that the State of Connecticut had been very helpful toward his work and that his dream is to ensure that every child is healthy. He mentioned he was distressed when the parents of a child with a treatable condition got turned away for lack of health insurance. He talked about meeting folks at the JCC and that as result he started receiving backing for his work from people including baseball player Johnny Damon.

Lesley Bennett stated that funding for much of the research starts with the support of organizations and patient advocates. She added that they seek to involve members of the community and businesses so that it is not all on the state to foot the bill.

Kristen Angel added that from NORD's perspective, they work very closely with non-profit organizations, and that they offer a research grant program. She added that anyone interested in raising funds for research can establish a fund through NORD and that when the money reaches a certain level it is released to the applicants. Kristen informed the group that NORD have had two therapies from their program get FDA approval.

Dr. Lutz asked Dr. Weinstein how hard was it to get FDA approval for clinical trials for the therapies he developed.

Dr. Weinstein stated that one the issues with Glycogen Storage Disorder is that there is a treatment but people have to be vigilant with their care. He added that when using the

corn starch therapy it must be administered to the patient every four hours including the night time. He stated that he had five children die because their parents overslept and missed the overnight dose. He added that it is a treatable disease but it is incredibly unforgiving as well.

John Hopper Presented on Fibrolamellar Cancer

**Insert presentation here.**

Dr. Lutz stated that from an oncology perspective sometime mouse models don't mimic human cancer very well and that the tumors and their growth are very different. She added that cancer has been cured a thousand times in mice but not once in humans and that she does have some mouse models where human tumors are transplanted for study. She stated that her lab does test many different types of cancer on her models and is always willing to try new experiments.

Adrienne Hoffman presented on her personal experience on being the parent of a child with a rare disease.

Dr. Gulati thanked Adrienne for sharing her very touching and moving personal story.

Dr. Gulati adjourned the meeting at 3:10 PM.