

**State of Connecticut**  
GENERAL ASSEMBLY



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

Meeting Summary

July 21, 2017

Dr. Gulati convened the meeting at 1:09 PM and asked Task Force members to introduce themselves.

Members Present: Dr. Cathleen Lutz, Dr. Joseph Tucker, Dr. Lynne Sherman, Dr. Mridu Gulati, Nancy Dupont, John Morthanos, Michele Spencer-Manzon, Lesley Bennett, and Sen. Terry Gerratana.

Others Present: Margaret May, Noah, Jacklyn Beirne, Julie Gortze.

Dr. Gulati stated that Dr. Spencer-Manzon would be presenting on Newborn Screening and John Morthanos will be discussing the Department of Public Health's (DPH) Oxygen website. She informed the group that the next meetings will be on September 8<sup>th</sup>, October 20<sup>th</sup> and November 17<sup>th</sup>. Dr. Gulati asked that Lynne Sherman, Maryann Mae, and Paul Pescatello speak at the September 8<sup>th</sup> meeting. She added that she has asked Marie Egan and Cat Lutz to present at the October meeting and that she will continue to reach out to various communities to participate in Task Force discussions. She reminded members that a report and recommendations of the taskforce is to be submitted to the General Assembly in January 2018.

Dr. Michele Spencer-Manzon stated that the task force should discuss what could be done to promote help for patients in the State of Connecticut. She said expanding the scope of Newborn Screening would be immensely helpful to patients and their families, who if diagnosed earlier on in life would lead to a better life with long term outcome. She added that the task force requires the support of state legislators to accomplish these tasks, which do not require much financially. Dr. Spencer-Manzon added that the state currently has some of the equipment that required for testing but the Legislature will need to first approve the equipment for use. She stated in the last year she has diagnosed six new cases of creatine disorders and that the youngest patient is six years old and the oldest patient is thirteen. Data coming out the state of Utah shows that treating kids at birth drastically improves the odds of them being developmentally normally. Starting treatment at age six or later as is done in the state of Connecticut can lead to mental

retardation. The state also needs to include storage disorders that are not currently tested for.

Dr. Spencer- Manzon stated that in Connecticut there have been two patients born with Pompeii disorder. She stated that she is treating the child and because his disorder was identified early enough, the child is now walking and off a ventilator. The other patient born with Pompeii that was identified late, about six months after has not done nearly as well as the first child and is not walking. She stated that she would like to see some of these rare diseases on newborn screening list in order to catch these disorders in their infancy. She again added that early treatment is effective and can save lives and should be a part of the proposal to General Assembly.

Dr. Joseph Tucker stated that it is important to think about support for families caring for these patients. He added that this support should include social work, financial support for medical foods, and medicine that are not covered by insurance companies.

Dr. Gulati asked where things stand currently as far as adding these rare diseases to the newborn screening list.

Dr. Michele Spencer-Manzon stated that some of these tests can be added to existing platform and the remainder of the tests needs to be part of a new platform that requires a special chip. She added the Connecticut General Assembly must tell hospitals to add it to them order for it to be tested.

Sen. Gerratana stated the first thing is to contact The Department of Public Health (DPH) to see how the test can be added to the existing protocol. She added that when doing legislation to implement new testing protocols the difficulty is that all new tests must be proven to be scientifically sound at the federal level and a cost analysis must be done at the state level. Sen. Gerratana added that she would be willing take the list to DPH to see if they can accommodate the request of the Taskforce.

Dr. Michele Spencer-Manzon stated she talked to DPH employees such as Adrienne Manning about adding these test to Connecticut newborn screening panel. She added these tests are already approved at the federal level and Connecticut simply has not implemented these them.

Dr. Gulati asked that Dr. Spencer-Manzon forward her work and findings to Sen. Gerratana via email so that she can review them and follow up with DPH.

Dr. Gulati asked if there are states doing test that are not currently approved by the federal government.

Lesley Bennett stated that one of the things that the rare disease community is looking for is that the state should pass legislation requiring hospitals to test for diseases within a year after they are appointed by the federal government.

Sen. Gerratana stated that she has done legislation in the past regarding rare diseases. She added that the proposed legislation to create this taskforce and is a good effort and she is glad the taskforce is meeting to discuss these important issues.

Dr. Gulati asked if there are other ongoing Taskforces/council etc. that are already making recommendations that overlap with this taskforce charge.

Sen. Gerratana explained that there are varying ranges of taskforces that have broad topics and that she has found the most productive ones tackle a particular area or concern. She provided an example where last year there was a taskforce on opioids. That gave an opportunity for legislators and members of the public to research, understand, and collect data on specific areas that may be of help to people in the state with these disorders. She added that the taskforce investigated how opioids are prescribed that came up with specific recommendations for the legislatures after consulting pharmacist and various other subject matter experts. She added the purpose of the taskforce is to help legislators come up with specific legislation that are based on current data and evidence based science on a particular subject matter. Sen. Gerratana urged that the taskforce to make recommendations that it is easy to digest that way. She stressed the importance of communication with legislators and the executive branch.

Dr. Gulati stated that one challenge for the taskforce is that the subject matter encompasses 7000 diseases and in terms of looking at some of the emerging themes it seems that other taskforces might have found similar themes as well. She added that the taskforce would be happy to work collaboratively with other taskforce and existing pieces of legislation to tackle the charge of the Rare Disease Taskforce.

Sen. Gerratana stated that if there is something really needs a remedy then the taskforce showed focus on the most pressing issues to start. She advised the taskforce to do things systematically in small segments, especially in light of the state's budget crisis.

Lesley Bennett thanked the Senator for attending the meeting and stated that support for families during newborn screening is incredibly important. She added that according to Dr. Zavosky at Department of Social Services (DSS) there is a three year waitlist once babies are identified and that must be discussed and further addressed. She further stated children with nutritional needs are cut off from funding when they turn thirteen and that she is aware of families that are practically going broke trying to feed their children and cannot afford the \$4000 a month cost for medical foods. Lesley Bennet stressed there needs to be ongoing discussions with the legislature because there are so many diseases. DPH is running into some issues with newborn screening and has asked herself and Dr. Gulati to get involved to offer insight.

Dr. Gulati introduced John Morthanos as he is going to present on the "Oxygen Link" DPH website Oxygen.

John Morthanos highlighted information on the DPH website and showed the fact it is not user friendly when looking for specific information regarding a rare diseases.

Dr. Gulati questioned the ability of a patient, caregiver or vendor to navigate the site. She further questioned if all the information about newborn screening is mapped out correctly and asked how we figure out who we could reach out to assist in developing the site.

Lesley Bennett reminded members that the group talked about committees and recommended that there should be four committees. One of those committees should be focused on education; one on caregivers and one on enhancing support for care givers and patients. She stated that cutting it down committee wise will allow the taskforce to focus on a few issues and make the case to Sen. Gerratana and the Public Health Committee because many of the issues needed help with are legislative. She added that they are insurance issues related to specialty tier drugs and many rare disease drugs that are really expensive and start at that tier.

Dr. Gulati recommended that a survey should be sent out and subcommittees and working groups be established. She listed newborn screening, DPH resources/website, durable medical equipment, palliative care services, and community professionals.

Lesley Bennett stated that it might be best if the Department of Education came in the month of October as the September meeting is already pretty full.

Dr. Gulati asked how parents communicate with their children's schools about their rare disease.

Dr. Tucker stated that it is different for each case. He added that in most cases it is not a problem and information is obtained from the parent. At times Dr. Tucker speaks with the school nurse directly and provides the nurse with written documentation if asked. It is important that parent give the physician permission to communicate with the school on their behalf and that if the parents do not allow that they themselves become the intermediate. He stated that most schools are very receptive of the students.

Margaret May mentioned that in the 504 Plan the treatment center nurses will go with the family to a meeting with the school and as the child gets older 504 Plan it is up to the family to make sure it remains active for the growing child. She added that the treatment center nurses are phenomenal about working with the schools talking with the staff about what is permissible and what is not. She further stated that having a 504 plan goes both ways and requires communication between all parties.

Dr. Gulati stated that there had been a school nurse task force and asked what areas they covered. She asked if specialty centers do education at the state, school, or individual level.

Joe Tucker stated that currently it happens with individual patients and usually Schools and nurses may do their own research at times to learn about conditions. He added that they may contact him with specific question and on a few rare cases he visited schools and that it all depends on the particular case.

Dr. Gulati asked what areas the Office of Early Childhood Development should cover when they come to the next meeting.

Lesley Bennett requested that the Office cover support programs for families with children diagnosis with a rare disease.

Dr. Tucker asked Dr. Gulati to clarify her question.

Dr. Gulati explained that she asked the Office of Early Childhood to address the taskforce on their overall experience caring for children of rare diseases. She added that the office asked her what areas and topics they should be prepared to speak on at the meeting. She asked the Office what the taskforce could do to help them and what areas they are for improvement.

Dr. Tucker stated that said there is always room for improvement for communication between the involved parties. Currently the burden for such communication disproportionately falls on parents. He added that in many cases this is adequate but if the

school is having difficulty he may get involved. He stated that nothing formal is set in place for that. Some parents use state advocates to get their children services.

Dr. Gulati questioned if folks who have issues navigating the system if there is a way to use technology to educate people and connect them resources and in particular school nurses.

Lesley Bennett asked Dr. Tucker how many patients of his have individualized 504 Plans.

Dr. Tucker stated many of his patients have various disabilities or difficulties. He does not have an exact number but almost all of the children have some type of special services in place with the school before they come to see him. That's often arranged by their primary care physician.

Lesley Bennett said it important that IEP be incorporated in those and that based on her experiences nurses are separate from the schools and unless the healthcare plan is in the IED of 504 plans often time administrators and teachers are not in the loop. Stamford Public schools started developing a packet of information for the parents and then had a person come in and educate the school. She stated that there are many issues still around that area. Something should be developed and implemented statewide.

Nancy Dupont thanked John Morthanos for his presentation and added that she is a frequent user of the site and can navigate it with ease. She also stated that she is aware of a developer that worked with DPH site to make the site more manageable. She asked if any of the other sites mentioned had a similar developer in mind.

Margaret May asked John Morthanos a technical question about the website. She went on to state the search engine of the website needs to be greatly improved and modernized.

Dr. Gulati adjourned the Task Force meeting at 2:23PM