New Hampshire Governor's Rare Disease Advisory Council
Meeting Minute 5/21/21
Prepared by Laura Landerman-Garber. Ph,D

-Chairman William Marsh read the NH State GUidelines for official NH meetings during the COVID-19 Pandemic, including the parameters for meetings held via teletechnology.

-Attendance:

Present: Chairman William Marsh, Co-Chairs Rep. Gary Woods and Dr. Mary Beth Dinulos, Dr. Sai Cheralas, Ms. Libby Shannon, Ms. Krista Gilbert, Dr. Elijah Stommel, Dr. Landerman-Garber. Absent: Senator Cindy Rosenwald, Drs. Lefleur and Shepard. Guests: Paula Minnehan and Courtney Tanner

-Minutes from last meeting read and approved without objection

-Database ALS Registry

Dr. Stommel had a phone conversation with a representative from the CDC, the Director of the National Registry. It was noted to be an encouraging discussion about the possible use of the National Registry for partnership with the development of a NH Registry.

Dr. Cherala raised some concerns such as costs, the need for it to go through the NH Legislature, and the issues related to privacy.

Dr. Stommel noted that patients could perhaps opt in or out and that some data-sharing could be limited.

Dr. Cherala noted that prior to getting the system up, it will need to be clarified by official privacy guidelines.

Chairman Marsh raised the question as to whether personal identification could be excluded from the reportable data and if that might not ease the limits posited above.

Rep. Woods expressed support overall and acknowledgement that the Legislators would need to clarify parameters.

Dr. Stommel raised concerns about how physicians/medical providers will get registry forms to patients who eventually go elsewhere, perhaps out of state, for treatment

Rep. Woods asked how many patients would get “lost” for example to a Boston treatment facility, who would then Dx the patient and the data would go to a Massachusetts registry.

Dr. Cherala noted that cancer Dx are often shared amongst varying states’ databases.

Dr. Stommel noted that if for example a NH patient goes to Massachusetts and become Dx there, that while the data for that patient will be tagged for Massachusetts (and not NH), it would be sent to the National Registry and the data overall would be in and could be used for the appropriate research, education, and treatment purposes.

Ms. Shannon discussed her involvement with the MG Registry and her connection with a woman in Massachusetts who perhaps works with that registry. She offered to contact her and connect her with Dr. Stommel.

Chairman Marsh noted that he, Rep. Woods, and Dr. Cherala might explore writing legislation if time allows and information is gathered for a possible filing in September. Ms. Tanner from DHMC offered to aid in any way that would be helpful. They will connect offline outside of the meeting and report back to the Council.

-Legislation

HB600 Newborn Screening passes in the Senate with one minor change

It was noted that Senator Rosenwald made a change related to a ‘no out of pocket expense for the patient’ inclusion

The Legislation will now return to Chairman Marsh and Rep. Woods Committee next.

-Youth Sudden Death

A synopsis of Dr. Dinulos’ discussion was graciously provided by her on 5/21/21 for inclusion here)

Dr. Dinulous attended the American College of Medical Genetics meeting (virtually) in April 2021. SHe reported on a presentation from Dr. Koch of the Boston's Children's Hospital group re: “ Genetic determinants of sudden unexpected deaths in pediatrics

(SUDP).” Over 10 % of infant and child deaths in the US occur suddenly, unexpectedly, and without cause. There are known extrinsic factors in SUDP , such as sleeping position, etc. but intrinsic factors, such as genetic predisposition, are not well known. The Boston group , along with the San Diego group from Rady Children’s Hospital, decided to take an “undiagnosed disease” approach to this issue by integrating comprehensive phenotyping and genetic analysis to evaluate the genetic predisposition in SUDP.They reviewed 352 cases of SUDP (ages newborn to 11) from 2012-2020 and found that 11-28% of of SUDP cases have an identifiable genetic variant that may account for, or add to, the child’s unexpected death. Testing performed was either whole exome sequencing (WES) or large panel testing that would encompass cardiac, neurologic, and systemic/syndromic genes. The majority of the mutations were cardiac or neurologic genes. The number of mutations found in the SUDP cases were much greater than those found in a control cohort. They concluded that genetic mutations should be a part of the SDUP evaluations for several reasons. First, it may provide a Dx for the parents who have just lost a child. Second, it would allow for medical surveillance for all at-risk surviving family members. And lastly, it would aide in recurrence risk counseling for the parents. These statistics are similar to other published studies, although not not a lot of data is available.

This information is important for requesting these types of genetic testings in NH SDUP cases.

Next steps will be to discuss this data with the NH Medical Examiners. We will need to know the number of cases per year that might warrant genetic testing. Dr. Cherala noted that in 2019 there were 124 child deaths (ages newborn to 21) and that 62 of them were “natural” deaths. We would then expect that a subset of those cases may SUDP. Once we know the number of cases that the ME feels would be appropriate for genetic testing per year, we can calculate the annual funding for that testing.

Rep. Woods asked what kind of testing would be warranted in these cases- WES or panel testing? Depending on the numbers and costs, it may be that large panel testing is the most cost effective route this time, but as technology changes, so will testing algorithms.

Dr. Cherala offered any PH information that would be useful in this regard.

Chairman Marsh inquired as to whether the existing statutes are sufficient or if expanded and or new legislation is needed. He offered to work with Dr. Dinulos in any way that would be of assistance.

Dr. Dinulos will speak to the ME about Dr Cherala's' questions about consent, especially in regard to genetic data.

-Naltrexone

Dr. Lefleur was absent and therefore the discussion that involves his research or views will be tabled until the next meeting.

Ms. Gilbert reported that the patients with whom she speaks continue to tell her that they are not able to get Naltrexone covered by their insurance. She expressed frustration about the delays of these patients not getting this and possibly other medications.

Ms. GIlbert did speak again with the Eastern State Pharmacy Staff and was informed again that it is difficult to get it covered as a compounded medicine.

Chairman Marsh reco that Ms. Gilbert and Dr. Lefleur meet to further discuss all of these and related issues. Chairman Marsh offered to participate with these offline discussions.

Ms. Minnehan asked Ms. Gilbert if she had any feedback from insurance companies and offered to be of assistance in this regard.

Dr. Cherala offered to inquirers with the Medicaid

Ms. GIlbert clarified that it is the LOW DOSE Naltrexone that is not covered and needs to be compounded.

Dr Cherala noted that low dose is considered an off-label issue and may be central to this concern

Dr Cherala inquired as to whether there is a list of conditions for which it would be used/helpful.

Ms. Gilbert was able to provide a list of medical conditions per Dr. Cherala's request.

Ms Gilbert believes that in the long term, Naltrexone will be produced in lower doses.

Chairman Marsh posited that there may be a need at some point for a legislative process. He reco that Ms. Gilbert contact Mr Gary Merchant

-Newborn Screening

Dr. Cherala noted that there is not an immediate need for discussion but there is an issue at hand; that being, if screening tests come back that are out of range and there is a need to go back to the pediatrician for more information, there appears to be some resistance. The message is that the legislation is not clear on guidelines

Dr. Cherala also inquired as to the current legislation that can be used regarding these needs for follow up

Dr. Dinulos inquired as to the source of the “push back”. Dr. Cherala posited that the concerns come from a risk management perspective and is exploring this with their legal team.

Chairman Marsh and Rep. Woods offered to be of assistance offline.

-Community Outreach

Ms. Shannon reported that she spoke with Ms. Gilbert about the ongoing limitation of COVID-19 in terms of reaching out to the RD Community. Ms., Shannon was not, for example, able to take flyers to doctor offices to share with patients.

Ms. Gilbert confirmed that RAN is currently not distributing flyers as well.

There was some discussion of possible community confusion about the different RD organizations, such as RDA, NORD, RDAC, and Rare Action Network and the need to clarify the different missions of each group.

Chairman Marsh reminded us that one of the charges is to raise community/ statewide awareness of rare diseases.

Dr. Landeraman- Garber and Ms. Shannon acknowledge the tabling of the creation of a facebook page but both feel that it may be an efficacious option in the future .

Dr. Dinulous inquired about the development of/creation of a website dedicated to the activities of the Council. Chairman Marsh noted that we do have a government webpage where our minutes and meeting times are posted. Dr. Landerman-Garber discussed her experience with website development for her nonprofit and the possible

gains to be made with it. She also noted the expense of such development. Future discussion on this issue will occur

-New Business

Dr. Cherala will be presenting our annual report on June 11, 2021

-The next Council Meeting is scheduled for June 18, 2021

-The Meeting was adjourned without objection.

Meeting minutes respectfully submitted by Dr. Landerman-Garber in May 27, 2021