

New Hampshire Rare Disease Advisory Council
Meeting Minutes 10/15/21

The meeting was held in-person with a Zoom option for those Council Members and guests who were not able or were not comfortable to meet in person secondary to COVID-19. Chairman William Marsh clarified to the attendees that because there was not a quorum present and that there would not be any voting throughout the course of the meeting.

Attendance

In-person: Chairman Representative William Marsh, Dr. Sai Cherala, Co-Chair Representative Gary Woods, and a guest - Jim Shannon

Via Zoom: Dr Angela Shepard, Dr. Laura Landerman-Garber, Ms. Krista Gilbert, Dr. Elijah Stommel, Co-Chair Dr. Mary Beth Dinulos, Sen. Cindy Rosenwald, Dr. Richard Lafleur, Ms Libby Shannon, and guests - Ms. Audry Gherkin, Ms. Abby Rogers, Ms. Courtney Tanner, Ms. Paula Minnehan

Minutes

Minutes from the previous meeting were reviewed. Approval of minutes could not be voted upon due to quorum limitations.

ALS Registry

Dr. Stommel reported that he spoke with an ally who is working at the national level to push legislation for mandatory reporting to the National ALS registry. He will follow the progress and report to the council.

Legislative Updates

Rep. Woods reported that he continues to collaborate with Rep. Rosemarie Rung on her proposed bill to establish a study commission related to the cyanobacteria in NH lakes. Rep. Woods reflected that he considered shifting energy from the ALS registry LSR in favor of focusing efforts on the cyanobacteria commission LSR. There was minimal discussion to support dropping the registry LSR. There was good support for continuing to pursue the cyanobacteria commission bill. Dr. Stommel offered that there are multiple health impacts as well as economic impacts related to the cyanobacteria in our waterways.

Rep. Marsh continues to press for legislation to allow the Rare Disease Advisory Council to meet remotely. He feels that this is a reasonable request given that many of the individuals we serve may be immunocompromised or have other significant health or mobility issues that make in-person meetings unsafe or impractical.

The Telehealth Bill was referred to interim study by the HHS committee. The Senate has a similar bill they are evaluating.

SDY

Dr. Dinulos said the SDY committee has not met since last month but that she had a conversation with the NH Medical Examiner. They discussed what samples are collected during autopsy, what genetic screening tests are available, and what number of pediatric cases might be appropriate for sending out additional screening. They determined that probably 30 cases per year might be appropriate and that a reasonable expected cost for genetic screening might be \$600 per panel. Therefore, the anticipated cost might be around \$50,000 yearly. Audrey Gherkin mentioned that we might look for grants to fund this if we move forward.

Newborn Screening

Dr. Cherala stated that the previous contract is up with the testing lab so the new RFP is out. They do not anticipate any disruption in service during the process. They have some new staff who will be collaborating with Dr. Shepard in some educational outreach she is doing for the NH Rare Disorders Association. Rep. Woods asked for clarification on the latest list of conditions included in the NBS panel. Dr. Cherala outlined the list and the process for how disorders are selected for inclusion.

Community Outreach

Ms. Gilbert reported that the Bureau of Developmental Services is working to redesign the waiver process and some families are concerned that there seems to be a bias towards residential care rather than accessing services in the community. The BDS will have meetings that will allow the public to voice their concerns and provide feedback on the process.

Ms. Gherkin shared that it is becoming increasingly difficult to find in-home assistance and direct service providers. Many families are unable to participate in the workforce or community because they must remain home as the sole care provider. Ms. Gilbert offered that we need to advocate for increased pay and standardized education for DSP's. Rep. Woods stated that there is a shortage of care providers in hospitals and nursing homes as well. There does not seem to be the necessary financial investment to draw in more workers.

Education

Dr. Shepard is trying to coordinate with Dr Vrooman on the LDN training for the NH Rare Disorders Association's education projects. As mentioned earlier, she is planning on developing some sessions on newborn screening with the help of DHHS staff. Hopefully NERGN will be interested in participating as well. She is developing some patient education offerings while work on the CME's is held up.

Next meeting will be held on November 19 and will again be at 29 Hazen Drive in Concord. A Zoom invitation will be sent out prior to the meeting for those unable to attend.

Respectfully submitted,

Angela Shepard, MD, MPH