Rare Disease Council Quarterly Meeting
Friday, November 17, 2023, 10:00 AM – 1:00 PM

Meeting Location:
All-virtual
https://www.zoomgov.com/webinar/register/WN_mdHPSdazT8KyQ2u6DqCYIg

Meeting Minutes

Council Member Attendance **Bold** = Present *Italicized* = Absent*	**Voting Record** Y=Yes, N=No, A=Abstain 5/16/23 Meeting Minutes
Voting Members	
Lisa Kaplowitz, MD, MSHA (Chair)	Y
Gwen Traficant (Vice Chair)	Y
Ijeoma Azubuko	Y
Wes Fisher	Y
Gregory Josephs	Y
Peter Kasson, MD, PhD	Y
Sharon Kopis, Ed.D. MS, RN, FNP- C, CNE	
Stephen Rich, PhD	Y
Elisabeth Scott	Y
Samantha Vergano, MD, FAAP, FACMG	Y
Stephen Green, MD, FIDSA	Y
Elissa Pierson	Y
Michael Friedlander, PhD	
Tiffany Kimbrough, MD, FAAP	Y
Angela Olmsted, PharmD BCPS	Y
Leslie Mehta	Y
John Feore	Y
Ex Officio Members	
Jennifer Macdonald, MPH, BSN, RN	
Samantha Hollins, PhD	
John Morgan, MD (Proxy: Rhonda Newsome)	
Vacant Seats	
Health Plan Companies Representative	

Virginia Department of Health Staff Attending: Christen Crews, Kelly Conatser

Council Business

- Christen Crews took roll call. Quorum achieved for meeting to proceed (16 of 21 members present)
- Each member of the council introduced themselves, including new co-chair, Gwen Traficant, introduced herself. Lives in Northern Virginia, teacher trained, husband diagnosed with rare liver disease, husband, daughter, and son all have had liver transplants. Currently works at Central Union Mission.
- Review of Agenda: Dr. Kaplowitz discussed requirements of RDC and meeting requirements and acknowledged challenge to achieve a quorum when meetings are held in-person. No additions made to the agenda.
- Meeting Minutes from May 2023: Gregory Josephs motioned to approve meeting minutes. Wes Fisher seconded the motion. Meeting minutes approved. (16 – Y, 0 – N)

Council Survey Options, Kelly Conatser, Virginia Department of Health

- A description of REDCap as a survey tool was given and capabilities were demonstrated. REDCap is a secure web based, fast/flexible, access through anywhere there is web access, customizable, data import/export feature (data collected through redcap), can generate survey PDF for offline use.
- Example of REDCap use: Title V uses REDcap, uses pictures/images/branching logic, captures both quantitative and qualitative data, shared images of different REDCap features. VDH Office of Family Health Services Epidemiologists can help support developing, distribute, and analyze data; disseminate PDF copy; draw conclusions based on survey results to drive decision-making.
- Rhonda Newsome asked if survey has the ability to translate into multiple languages. Kelly Conatser stated it did.
- Funding (lack of) has prohibited large scale dissemination. REDCap is a practical way to start survey efforts. Christen Crews talked about equitable survey distribution need to mail as well, as discussed previously with starting electronically and then using pilot data to help secure funding for fiscal impact support of larger dissemination.
- Dr. Kasson stated that the use of REDCap is highly aligned with council's strategy.

Group Survey Discussion and Next Steps, Christen Crews, Virginia Department of Health

- A survey was disseminated to Council members in September to identify key priority areas and what to focus on. High Priority topics should take about 10 minutes to complete. Low priority topics would have the option to expand it to contribute more information. Target audience would be those with Rare Diseases and parents of those with rare diseases. Christen Crews shared the responses of the survey: Six topics were noted as high priority topics, "Knowledge and Support" was highest.
- Interactive Poll conducted reviewing topics of survey and potential questions related to each topic. Members stated the sequencing of the questions will be important. Christen Crews explained that the questions presented today are grouped by topic area. Questions will be optional to respond (not required) to maximize survey responses. Additional potential questions around topics included:
    - Demographics: add 5 digit zip code for location of respondent.
    - knowledge and support
    - rare disease
    - Priorities of the Virginia RDC
    - health follow up and diagnosis: For question 5 & 6 could there be examples given? Members expressed concern about survey fatigue if there is too much to read, and to leave it open ended.
    - insurance: capture denials, combine questions 3-5: if denied coverage, use branching logic to include reasons why denied (multiple options) and open text for "other"
    - specialist centers/care: add out of state as option
    - patient satisfaction

- PROMIS 29*
- pediatrics
- PROMIS 29*
- qualitative feedback
- relocation: check for redundancy with insurance, potentially combine questions but add distance to insurance.
- stigma scale
- anticipated stigma scale

- Council suggested adding questions about financial/emotional impact on caregivers and support. Co-chair Gwen Traficant will compose 2-3 questions and send to Christen Crews so she can add to the questions to be sent to Council.
- RDC Support Staff will send draft survey to Council members to review, and pilot distribution will go to certain individuals for review and feedback before dissemination plan (to be determined).

Public Comment

- Ms. Kelly Danoy – Last week she shared public comments with Virginia NBS AC as well. Her daughter has Krabbe Disease (early infantile onset). 2015 and 2020 Krabbe disease was not recommended to be added to the NBS panel. A timely diagnosis would increase supportive care for her daughter. The Council has the ability to influence policy and supportive care to those infants and families diagnosed with Krabbe and other rare diseases.

Supporting PCPs to Care for Rare Disease Patients – Dr. Regier, Children's National Hospital

- 1 in 10 are diagnosed with a rare disease, over 7,000 rare disease
- NBS is a good way to diagnose rare disease but how else can rare diseases be diagnosed and found?
- Building up the rare disease clinical community – rare disease clinical research program (year long course)
- Expansion into disaster science course launching in 2024 – to aid in rare disease needs in disasters
- Non-Rare Disease Clinician support – GeneClips is an app that shares genetic information on rare diseases (both clinicians and those with rare diseases)
- 4 hour CME program with four education areas: testing, culturally competent care, resources and how to care for yourself. Shred learner engagement and demographics of those taken the course so far.
- Demonstrated RareCAP that launched in October 2023. – clinical care protocols for rare disease patient care.

Council Business

- 2023 Annual Report was submitted to the Commissioner and SHHR and in review.
- Survey will be sent out for 2024 Council meeting dates. Please poll for alternative locations.
- NORD Summitt and Rare Disease Advisory Council meeting updates provided by Dr. Kaplowitz. Many states are in the stage that Virginia is in (councils). Breakouts were very specific: FDA perspectives, orphan product development, clinical studies, gene therapy, etc.
- Rare Disease Fund document is in development and review. Will have draft at next meeting.
- Action items for next meeting: Survey draft questions sent for final review, Gwen to compose a couple of caregiver questions. Initial talk of dissemination plan. Suggestions for speakers.
- Next meeting will be in person.
- Meeting adjourned at 1:09pm