



**North Carolina Rare Disease Advisory Council Meeting**  
**Tuesday, June 4<sup>th</sup> 2019**  
**The Frontier**  
**800 Park Offices Drive**  
**Research Triangle Park, 27709**

1:15 – 1:20	Welcome, Introductions of Guests	Tara Britt
1:30 – 1:45	Legislative Updates & Discussion on NC Rare Disease Networks	Rufus Edmisten Tara Britt
1:45 – 1:55	Patient Advocacy Story	Colin Connors, IDF Health Patient Advocate
1:55 – 2:10	Gene Therapy Updates	Aravind Asokan
2:10 – 2:20	NC Medical Genetics Association Meeting	Stephen Lehrman
2:20 – 2:35	Patient Advocacy & ROI Updates	Sharon King Mike Knowles
2:40 – 3:10	ACMGG “Current Conditions in Medical Genetics Practice” & State Plan Group Updates	Stephen Lehrman Kerry McCarthy Adams

Questions? Tara Britt, 919-417-6130  
Minutes

## Meeting Attendees:

Cindy Powell (UNC)  
Ali Smyth (IQVIA)  
Ray Huml (Patient Advocate , MDA & IQVIA)  
Brenda Nielsen  
Kerry McCarthy Adams (Co Chair NC State Public Health Plan for Genetics & Genomics)  
Sandi Lehrman (Patient Advocate & RD Advisory Council Task Force)  
Stephen Lehrman (Patient Advocate and RD Advisory Council Task Force)  
Jenny Klein (Patient Advocate, MPS)  
Rufus Edmisten  
Becky Sansbury (Patient Advocate &  
Mike McBrierty (Biogen)  
Colin Connors (IDF advocate)  
Sharon King  
Mike Knowles  
Mary Ann Naegle (Centogene)  
Lee Ann Lawson (Amicus)  
Tara Britt  
Vandana Shashi

Rufus and Tara gave legislative updates regarding the Council budget. Rufus mentioned our meetings with Representative's Brisson & Carney. We also discussed the meeting with Senator Woodard, Chair of Life Sciences Caucus. Rufus, Stephen and Tara attended that meeting.

Colin Connors representing the Immune Deficiency Foundation (IDF) was our guest patient advocate speaker. There are more than 350 immune deficiency disorders. Colin spoke about his family dealing with one of these immunodeficiency diseases. The IDF mascot is Think Zebra because in medical school, many doctors learn the saying, "when you hear hoof beats, think horses, not zebras" and are taught to focus on the likeliest possibilities when making a diagnosis, not the unusual ones. However, sometimes physicians need to look for a zebra. Open discussion about think zebra when we are all looked at as horses and how newborn screening really does save lives.

Mike Knowles asked if we should have a rare disease caucus within NC legislature. Mike McBrierty added that the Life Sciences Caucus was very engaged. Stephen Lehrman also discussed the importance of getting NC BIO involved. Mike McBrierty also talked about the monthly Life Science Caucus breakfast meetings and that it would be good for the council to speak at one of those meetings. Sandi mentioned it would be great to have a rare disease caucus with so much going on in Life Sciences.

Cindy Powell gave us some state lab updates in absence of Mark Benton. She and Kerry also discussed NBS and NC DHHS priorities to get Pompe Disease, MPS-I, and X-ALD approved last year included in the State NBS screening panel. Cindy mentioned that the federal Newborn Screening Saves Lives Reauthorization Act of 2019 needs to pass Congress in order to reauthorize the Secretary of HHS' Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC).

Stephen talked briefly about the Council's report on Access to Genetic Testing and a recent paper by the American College of Medical Genetics and Genomics (ACMGG). The paper provided results of a 2015 survey of medical genetics providers and validates most of the findings in the Council's report. Stephen also said that the cost of genome sequencing had followed Moore's Law up until 2007. Since then the cost has dropped by a factor of 10,000 times.

Sandi talked about two meetings at the federal level. The NCATS meeting and UC Berkeley forum meetings. The NCATS meeting took place on June 11<sup>th</sup>, but see the following for agenda if you want to follow up. [https://events-support.com/events/NCATS\\_CNS\\_AAV\\_Immunogenicity\\_Workshop/](https://events-support.com/events/NCATS_CNS_AAV_Immunogenicity_Workshop/). Tara is going to follow up with PJ Brooks regarding outcomes and will distribute.

More about the UC Berkeley Forum for Collaborative Research can be found here: <https://hivforum.org/projects/rare-diseases-forum/rare-diseases-forum-meetings>  
If your company is interested in becoming a member, you can email Veronica Miller, [veronicam@berkeley.edu](mailto:veronicam@berkeley.edu) and Brenda Rodriguez, [brodriguez@forumresearch.org](mailto:brodriguez@forumresearch.org).

Ray Huml and group discussed costs of rare disease.

Mary Ann Naegle, Ray and Ali discussed payers.

Sharon King talked about the patient advocacy portal and the 7 patient advocacy meetings across the state. Sharon talked about these patient sharing stories and discussed the 5 pillars (diagnosis, expert care, research, community support, and access to therapies). She also discussed the quality of life for patients and model of value centered care.

Sandi talked about workforce development and Stephen also added the NC BIO Invest in Cures meeting scheduled for September 25, 2019.

Tara discussed and is coordinating the rare disease networks across the state and with those meetings with healthcare systems employees, researchers, clinicians and community based healthcare groups, Sharon should start those meetings with patient feedback she has collected.

Mike also gave a brief update on his portion of the ROI.

Kerry Adams discussed the NC State Public Health Plan for Genetics & Genomics. She and Stephen talked about the Task Force Genetic Testing report and how it aligns with the state's efforts.

Meeting adjourned at 3:30