



North Carolina Rare Disease Advisory Council Meeting
Thursday, June 28, 2018
Adams Building, NC DHHS Campus

1:00-1:15	Welcome & DHHS updates	Bruce Cairns Mark Benton
1:15-1:25	Council Activities/Legislative Updates	Tara Britt Rufus Edmisten
1:25-1:50	Patient Advocacy & Task Force discussion Advisory Council	Sharon King Stephen and Sandi Lehrman
1:50-2:10	ROI Patient Portal Update	Sharon King, Van Daughtry & Mike Knowles
2:10-2:20	Guest Speaker Metabolon	Mike Milburn
2:20-2:45	Research Opportunities	Bruce Cairns Mike Knowles Vandana Shashi
2:45-3:00	Future Objectives and Strategies	All

Questions? Tara Britt, 919-417-6130

Attendees

Council Members

Brenda Nielsen
Bruce Cairns
Mark Benton
Rufus Edmisten
Mike Knowles
Sharon King
Aravind Asokan

Guests

Nicole (Taylor's Tale intern)
Bryon Scott
Mike McBrierty
Mike Milburn
Laura Gunter
Susan Nichols
Becky Sansbury
Cindy Powell
Van Daughtry
Sandi & Stephen Lehrman (via phone)
Vandana Shashi (via phone)

Dr. Bruce Cairns, Chair and Mark Benton, NC DHHS, opened the meeting, welcomed group. Introductions were next.

Mark provided updates with newborn screening legislation and NC DHHS updates.

Rufus Edmisten & Tara Britt discussed legislative update regarding the appointment extensions for council members to 2023 and also about creating the rare disease networks across the state.

Stephen & Dr. Sandi Lehrman, Task Force, proposed the task force's next project to submit to the Advisory Council. Genetic Testing for Rare Diseases. Sharon also expressed what the Patient Coalition would like the council to address.

Discussion regarding who the audience is for the Task Force reports, General Assembly and Advocacy groups included. Dr. Cindy Powell (UNC) discussed that labs could work with patient insurers. Companies won't work with NC Medicaid. Bruce mentioned we need thoughtful guidance considering this. Vandana also brought up the need for genome sequencing.

Bruce asked for a vote from the Council regarding authorizing the Task Force to start working on the Genetic Testing/Genome Sequencing report. The Council unanimously agreed for the task force to submit a report regarding genome and exome sequencing.

Rufus asked to clarify what the 5-7k does for sequencing. Bruce asked Task force to educate the council regarding all of this. Include basics of Genetics in the report.

Mark Benton would like to offer assistance from NC DHHS in resources. Oregon Health Sciences and Washington University.

Sharon talked about the Coalition and how it predated the Council. She talked about the coalition having representation from all over the state, connecting with the community. Discussed the focus groups for ROI project.

Van Daughtry discussed the ROI and the patient portal. Patients driving. Patient centric.

Mike Knowles talking about his ROI project. Talked about Primary Ciliary Dyskinesia. He discussed the problem in diagnosis with this disease. Fast forward 15 years, identified genes that causes this. Genetic technology moving quickly. Mike talked about in finding one disease, other diseases have been identified.

Bruce talked about grant funding with CTSA and NIH grant funding.

Mike Milburn from Metabolon was the invited guest speaker. Mike talked about Metabolon and the company focus. Gave overview of the company. New technology that would help accelerate diagnosis for rare disease.

Cindy mentioned UNC is using Metabolon. Bruce also wanted to see how Metabolon could work with Burn patients and how do we bring all of this together.

Craft the wording of how this could work together in context so physicians and scientists could work efficiently.

We need to know how to answer some of these questions. Where is Metabolomics going in 3-5 years? Take complex and chaotic topics and put it into a strategy for the state to understand.

Aravind talked about Gene Therapy field. Explosion in field of Gene Therapy in the next several years. Great advances in SMA, DMD, and a type of rare eye diseases, due to single genes in rare disorders. 70 different trials in Gene Therapy going on. Identifying genes underlying key to these successes. Aravind and council discussed Gene editing and how important that is so you are precision engineering the gene.

TAG Therapeutic Approaches to Genetics Diseases. Diagnosis Mechanism to Therapeutics.

Mark talked about Newborn Screening (NBS). Process of identifying equipment, personnel for the 3 new tests. Legislation provides for ongoing financial to support this. New funding will promote longevity for NBS program.

Bruce thanked everyone for attending & closed meeting.

