



North Carolina Rare Disease Advisory Council Meeting
Thursday, February 22, 2018
Adams Building, NC DHHS Campus

9:00 – 9:10	Welcome & DHHS updates	Bruce Cairns Mark Benton
9:10 – 9:20	Introduction of Guests & Overview of Council	Tara Britt Bruce Cairns
9:20 – 9:35	Council Activities	Rufus Edmisten Tara Britt
9:35 – 10:10	Patient Advocacy & Task Force discussion	Sharon King Stephen and Sandi Lehrman Advisory Council
10:10 – 10:25	Legislative meetings	Rufus Edmisten
10:25 – 10:40	Research Opportunities	Bruce Cairns Mike Knowles Vandana Shashi Aravind Asokan
10:40 – 11:00	Future Objectives and Strategies	All

Questions? Tara Britt, 919-417-6130

Minutes from 2/22/18 Council Meeting

Attendees:

Council Members

Mark Benton, Deputy Secretary, DHHS

Rufus Edmisten

Bruce Cairns, Chair

Vandana Shashi

Sharon King

Brenda Nielsen

Mike Knowles

Tara Britt, Associate Chair

Guests

Pasha Donmez, Project Manager for Council and NC RDII activities

Yannis Viniotis, Professor, NCSU and Rare Disease Researcher and Advocate

Susan Nichols, CEO, Falcoln Therapeutics

Erin Frey, Senior Director, Baebies

Mike McBrierty, Gov't Liaison, Biogen

Laura Gunter, Membership Director, NC BIO

Stephen and Sandi Lehrman, Task Force Directors & Patient Advocates

Sandra Hall, Patient Advocate

Bruce Cairns, Chair, opened the meeting. Following introductions, Dr. Cairns reviewed the Council's purpose.

Mark Benton, Deputy Secretary, NC DHHS, discussed the department's focus areas and statelab shortfall of funds.

Sandra Hall, patient advocate, shared her personal story of her daughter's diagnosis with Spinal Muscular Atrophy and participation in a clinical trial. Mrs. Hall stressed the importance and benefits of Newborn Screening (NBS).

Sharon King discussed NC Rare Disease Coalition participation in the development of a Patient Information Portal through the GA ROI grant.

Stephen Lehrman gave an overview of the Task Force report: **Newborn Screening and Early Intervention for the Treatment of Rare Diseases – A Win-Win for Children with Rare Diseases and North Carolina**. The Council ratified the report of the Task Force.

Dr. Vandana Shashi shared an overview of the NIH Undiagnosed Network at Duke. Dr. Shashi and Dr. Mike Knowles led a discussion on the need for improved diagnostic services in NC and the opportunity to invest in genomic testing to find treatments.

Dr. Knowles described the NIH PCD Network that he leads at UNC, the importance of the ROI grant and the use of electronic medical records to diagnose diseases. Great progress

can be made in the next 5-10 years due to the screening of thousands of rare diseases. He stressed education as an important first step.

Dr. Cairns explained the Council's interface with the NIH and CTSA's and the ability to create partnerships with other state entities.

Deputy Sec. Benton stressed the importance of identifying treatment and suggested the Council visit the State Lab.

All agreed: Early diagnosis is key.

Rufus Edmisten & Tara Britt discussed meetings with Sen. Pate & Rep. Carney re a budget to support Council activities, including support for building a Rare Disease Community Network across NC.

The meeting adjourned at 11:00 a.m.

Followup: Sec. Benton, Rufus Edmisten and Tara Britt -- NBS