

North Carolina Rare Disease Advisory Council
Virtual Meeting
March 29, 2021

Attendees: Dr. Bruce Cairns, Rufus Edmiston, Sharon King, Dr. Michael Knowles, Stephen Lehrman, Dr. Sandra Lehrman, Dr. Cindy Powell, and Tara Britt.

Dr. Bruce Cairns called the meeting to order at 3:03 pm.

1. Golin Kosowitz, Patient Advocate, Brandon's Battle Foundation for SKS Kids (Smith-Kingsmore Syndrome). Sharon King introduce Golin Kosowitz of Charlotte, NC whose son Brandon is diagnosed with Smith-Kingsmore Syndrome (SKS). Golin and Aron Kosowitz founded Brandon's Battle Foundation for SKS Kids (<https://www.bbfskskids.org/>) to support research on treatments for SKS. SKS is an ultrarare neurological disorder caused by mutations in the MTOR gene and affecting about 100 children and characterized by a head that is larger than normal (macrocephaly), intellectual disability, and seizures. Mrs. Kosowitz said that until January 2021 Brandon's symptoms were mild. However, on January 18 Brandon was found unresponsive and was admitted to Levine Children's Hospital in Charlotte where he has been for the past 2 months. Brandon's Battle Foundation for SKS Kids has raised \$100,000 for research. The Foundation is working with Dr. Tony Hickey and the Catalyst for Rare Diseases at UNC on repurposing FDA approved drugs for treating SKS.

2. Legislative Updates

- a. **North Carolina Newborn Screening Update** – Tara Britt reported that she received the attached update from NC State Laboratory of Public Health Director Dr. Scott Shone. Dr. Shone said that NC should begin newborn screening for SMA within the next 6 weeks. Dr. Shone also said that newborn screening for X-ALD, MPS I, and Pompe disease will be implemented in 2021.
- b. **Federal Legislation** – Tara Britt reported that the Rare Disease Innovation Institute (RDII) continues to work with a federal steering committee including representatives from NCATS to make recommendations for creating International Classification of Disease (ICD) codes for each rare disease. Congressman Butterfield is a co-chair of the steering committee which is planning a workshop for 2021. Tara and Everylife Foundation spoke with Senator Burr's staff regarding co-chairing the steering committee and reauthorization of the Newborn Screening Saves Lives Reauthorization Act of 2021.
- c. **Extension of Appointments** – Rufus Edmiston and Tara Britt reported that they are preparing a request to the NC General Assembly to extend the terms of the Advisory Council members.
- d. **Other** – Tara Britt reported that RDII, Neena Nizar, and Khrystal Davis (Texas Rare Alliance) will participate in a call organized by PJ Brooks (NCATS) to discuss forming an NIH Rare Diseases Institute.

3. Expanding Representation of Advisory Council – Sharon King discussed the importance of broader geographical representation from academic/medical institutions in NC. As an example of the changing medical school landscape, Sharon said Atrium Health and Wake Forest Baptist Hospital recently announced plans to open a second campus for the Wake Forest School of Medicine in midtown Charlotte. The consensus of the Advisory Council members was that the Advisory Council membership not be expanded but that representatives of medical schools, academic institutions, rare disease advocacy groups, and industry are welcome to serve as advisors to the Advisory Council.

4. **Project Baby Owl** – Dr. Cindy Powell and Tara Britt, representing RDII, discussed conversations with Dr. Stephen Kingsmore, President and CEO, Rady Children’s Institute for Genomic Medicine, regarding the Project Baby Bear pilot project in California to use rapid Whole Genome Sequencing (rWGS) to diagnose critically ill newborn children. Dr. Powell said Project Baby Bear was funded by the State of California and that a similar pilot project in NC would probably seek state funding. Dr. Bruce Cairns said the Advisory Council could help assess the benefits and costs of rWGS for NC newborns but that the Advisory Council typically does not advocate for specific projects. The Advisory Council asked Dr. Powell and Tara Britt to keep it informed of their discussions with Dr. Kingsmore.

The meeting adjourned at 4:52 pm. The next meeting of the RDAC will be Monday, June 28, 2021 at 3 pm.