

Rare Diseases Advisory Council

July 26, 2024

Meeting Minutes

1. Welcome
2. Introductions
3. Newborn Screening Protocols presentation by HCU Network America Communications Manager Liz Carter
 - a. **Goal of HCU NA is to get the methionine level at birth cutoffs lowered from 65 to reduce the likelihood of missing a diagnosis of classical HCU at birth.**
 - b. **HCU NA has an upcoming meeting with the SC NBS lab meeting to discuss recommended newborn screening recommendations and new assays discovered by the CDC for improving HCU detection (Dr. Chapaigne was invited to join).**
 - c. **Neena Champaigne discussed her time and efforts on the SC NBS lab advisory council.**
 - i. **Explained the importance of getting on having concrete numbers reflecting at birth methionine levels and delayed diagnosis.**
 - ii. **Explained potential risks of lowering the methionine levels too much (ex. Typically raised levels in premie population), but also suggested that there were potential tiered testing solutions that should be explored.**
 - iii. **Expressed interest in future inclusion in HCU NA's interactions and efforts with SC PHL**
 - d. **Council follow-up items: Determining what action items requested by Liz that the RDAC can/wants to pursue:**
 - i. ***SCRDAC Considerations***
 1. **Would you consider including a question re: missed NBS cases in SC on your Rare Disease Needs Assessment?**
 2. **Would you consider putting an excerpt on your website or in a newsletter from HCUNA re: this issue?**
 3. **Would you consider communicating a recommendation on the need to review NBS protocols for HCU to your state NBS program?**
 - ii. **Patrick Flume suggested that we invite someone from NBS lab to join and SC RDAC meeting to explain their process and decision-making workflow**
4. SC RDAC Charter
 - a. **The council supports Dr. Flume's efforts to have the terms of council members changed.**
 - b. **Ginie Daguisse suggested having both 2 and 3-year terms so that there is not a loss of institutional memory**

- c. Karen Kemper suggested that after the initial term, the council could renew members for up to an additional set number of terms. She explained that this has worked well for other advisory groups of which she has been a part.
- d. Jonathan Hawayek said that some councils have an initial term and then, with vote from the council, a member’s term could be extended to a different amount of time.
- e. Jonathan Hawayek suggests we be cautious in bringing on too many people to the council, as it could become too difficult to manage. He suggests bringing people on an ad hoc basis or the use of subcommittees to leverage the expertise of certain individuals depending on council initiatives.
- f. Some states do not allow for the RDACs to lobby/testify for legislation or offer letters of support for initiatives. When developing the SC RDAC charter, we should confirm that we are not limited in that way.
- g. Dr. Flume suggested we consider adding a new section to the charter, under the council’s responsibilities, outlining the ways in which the RDAC might offer recommendations to the state.
- h. **Council follow-up items:**
 - i. **Dr. Flume to look into getting permission to edit council terms of service**
 - ii. **Dr. Flume to confirm that we aren’t limited in ways we can lobby or offer support for initiatives.**

5. Housekeeping and Additional Topics

- a. Recommendation for Rare Disease Organization representation
- b. SC-RDAC Progress Report was submitted to the SC Governor
- c. SC Rare Disease website updates (scrareddisease.org)
- d. **The council asked for an update on the needs assessment.**
- e. **Council follow-up items:**
 - i. **Jonathan Hawayek to contact NORD for potential recommendations on who to fill our RDO council spot.**
 - ii. **Jonathan Hawayek will provide codes for prevalence and incidence data available for 1 through 500 to Ginie Daguise to take them to get epi data from the state.**
 - iii. **Jonathan Hawayek will find contact information for Minnesota RDAC needs assessment so we can invite them to meet with us to discuss their findings and the development and distribution of the survey.**

6. Adjourn

Membership Attendance:

Representation	Member	Presence
Chair	Patrick Flume, MD (MUSC)	P
Department of Health and Environmental Control	Ginie Daguise (SCDHEC)	P
Department of Health and Human Services	Kevin Wessinger (SCDHHS)	A

Medical University of South Carolina	Dan Judge, MD (MUSC, cardiology)	A
Prisma Health -University of South Carolina	Divya Ahuja, MD, MRCP (Infectious Diseases)	A
South Carolina Hospital Association	Kate Wink (Santee Cooper)	A
South Carolina Primary Healthcare Association	Vicki Young (SCPHCA)	A
Biopharma Industry	Jonathan Hawayek (SPARK Therapeutics)	P
Research and Treatment of Rare Diseases (3)	Maysen Mesaros, MS (MUSC, neuroscience)	A
	Neena Champaigne, MD (Pediatric Genetics)	P
	Chip Norris, PhD (MUSC, connective tissue)	A
Patient (2)	Anthony Hamilton (familial TTR amyloid)	A
	Karen Kemper, PhD (scleroderma)	P
Rare Disease Organization	N/A	
Caregiver of person with rare disease	Jason Smith (hypoplastic left heart syndrome)	A
State Health Plan	Tripp Jennings, MD (VP and Clinical Innovations Officer, BCBS South Carolina)	P

Non Member Attendance: Liz Carter (HCU Network America), Danae Bartke (HCU Network America), Holly Murphy, Kevin Mann (Novo Nordisk), Sara Stolfus (Artia Solutions), Samantha Darwak, Tara Tsagouris (Jazz Pharma), Kemi Olabisi (Acadia Pharmaceuticals)