Tennessee Rare Disease Advisory Committee (RDAC)

Our mission is to improve the quality of life of individuals affected by rare diseases in Tennessee through collaboration, education, support and advocacy.

Minutes August 25th, 2021

RDAC Members In Attendance: Scott Strome, RDAC chair, Terry Jo Bichell, RDAC Vice Chair, Gillian Hooker, RDAC secretary, Rizwan Hamid, Suzanne Rock, Kim Stephens, Megan Crow, Lora Underwood, Regional French, Chip Chambers

Others in Attendance: Kate Segal, Sarepta, Todd Barber, UTHSC, Abby Trotter, LifeScienceTN

Agenda Item	***Notes***	***Action Items***
Rare Disease Patient Perspective	**Alexis Gordon** Currently writing an article about sickle cell disease and shared from the article she is working on. Article titled "Blessings and Miracles" Faith, education and self-advocacy She has Thalassemia beta plus sickle cell. When she transitioned to college, she had a care plan, and has had to maintain a care plan throughout her life. She now gives back to the sickle cell community through her many, many endeavors with the state Sickle Cell Foundation. Q: What has been your experience as a leader in the sickle cell community, what are the challenges for the community to get access to new treatments. A; Hydroxyurea is a main treatment, that helps with sickling of cells, helps prevent pain episodes. Also there are several new drugs, most of the drugs are too expensive for sickle cell patients to afford (as much as $10-12K/pill). These are not realistic costs. Also, the CRISPR therapies - she's hearing a lot of success stories. Finally, some patients get bone marrow transplants, which is harrowing - both to qualify, to find a match, intense daily medical care Q: To Tennessee specifically, what are the challenges and opportunities. She was able to find a team of doctors, hematologists, primary care, well versed in her disease here. That's been critically important. In the rural counties, especially out east, it can be more challenging Q: How was she diagnosed? She was born right after mandated testing in the early 70s to a mom who	

	was a nurse. Being a nurse, her mom was "in the know" and set her up to be an outlier. She was first qualified as "trait" then officially diagnosed with sickle cell disease in late teens, then in young adult years, she was identified as having beta thal plus. Q: Can you describe the episodes of pain? She describes her pain episodes as muscle spasms x 100. Mainly in joints, because that's where blood collects, deprives that body part of oxygen. If she catches an episode early enough - drink water, rest, it will only last one or two days. Otherwise it can last 4-6 days. For treatment, includes, rest, oxygen, hydration, OTC pain meds, hydrocodone, if unbearable, have to go to opioids.	
Sunset Hearing w/ TN Comptroller	**Gillian Hooker, RDAC Member** **Terry Jo Bichell, RDAC member** RDAC Sunset hearing is coming up in September. Terry Jo Bichell and Gillian Hooker will attend in Nashville - September 14th @ 1 pm. They have sent a list of questions over for us to address, Terry Jo will circulate a document with a draft of the response to the questions. We can come in with a 10 min presentation. It may not happen right at 1 pm. Include a slide about where we are going.	Terry Jo Bichell and Gillian Hooker will complete report and draft slide set. Abby Trotter will go back to original sponsors and let them know about the review, share the quick report with them.
Annual Report Development	**Gillian Hooker, RDAC Member** **Terry Jo Bichell, RDAC member** Report will summarize learnings of the RDAC over the year. The survey, our guest speakers, our initiatives. Will also lay out plans for next year, and beyond. Discussed three part structure of the report: 1) Diagnostics 2) Treatment 3) Innovation Conversations about dissemination. How do we best do this? Look at what is in this report. Getting the letter out to key stakeholders about what we're doing. Those legislators who supported our bill, key legislators in healthcare in TN.	Committee will begin drafting the report. Members: Terry Jo Bichell, Gillian Hooker, Abby Trotter, Rizwan Hamid, Kim Stephens
TN Expanded Genomic Diagnostics Program update / TN Rare and Undiagnosed Disease Network	**Rizwan Hamid, RDAC Member** **Scott Strome, RDAC Member** **Terry Jo Bichell, RDAC Member** Presented a set of slides, with a draft proposal. <u>Core problem</u> - Lack of access to genomic testing - Lack of access to rare disease specialists The purpose of this initiative is to identify the causes of rare and undiagnosed diseases and bring answers to afflicted individuals in TN	Terry Jo will lead the scheduling of the next meeting of this group.

	hope and ways to improve their health. **Overall Structure** Create a TN Rare & undiagnosed disease board, 3-5 individuals, report regularly back to RDAC- oversee Rare & Undiagnosed Disease Network - the doctors/teams doing this. Three scenarios of how this might function: 1) PCP or specialist orders genomic sequencing (RDAC removes barriers to precert and explains coverage criteria for all health plans- build a standard for coverage in the state) - results returned - diagnosis obtained - local team manages patient or refers out to rare and undiagnosed network? 2) Private insurance will not cover genomic sequencing. Could we find financing for people who do not have coverage? 3) PCP or specialist suspects a rare disease but has no idea what to do - they could present the case to the network, the case could be discussed. RDAC will work with health plans to establish criteria for WES and WGS, Establish funds to cover genetic sequencing if medically appropriate but not covered by a health plan. Fund a professional at each institution to coordinate the project. Q: Costs of sequencing -could this be offset w/ research potential. Q: What if it's a clear rare disease diagnosis and/or what if there is no result identified? Q: How would genetic counselors be involved? Question to be discussed in follow up meeting.	
TennCare Legislative Update - S.B. 1617	**Ashley Reed, TennCare** Ashley Reed provided an update on S. 1617, Public chapter 569.	
Pharmacy Advisory Committee Update & Planning for upcoming meeting	**Reginald French, RDAC Member** **Lora Underwood, RDAC Member** The PAC met on 8/12, there were 12 drugs presented, 1 related to rare disease - Kesimta for MS. Minutes will be posted to the website in the next couple of weeks. Next PAC meeting is in November.	
DUR Update	**Megan Crow, RDAC Member** Next meeting is in October	
Newborn Screening / Genetics Advisory Committee Updates	**Rizwan Hamid, RDAC Member** Will update on the next call.	

Department of Intellectual and Developmental Disabilities	**Terry Jo Bichell, RDAC Vice Chair** The statewide committee met two weeks ago. It was a very well organized hybrid meeting, with some in-person and some on zoom (for future reference, that made it very difficult to participate for those who were not in-person). The council really works intimately with the DIDD group and reviews and supports programs of the agency and other relevant TN programs. Minutes from recent meetings will be circulated to RDAC members for further background.	
Survey Update	**Kim Stephens, RDAC Member** Survey Updates were circulated to the RDAC members by email.	All RDAC members are asked to continue recruiting individuals impacted by rare disease to complete the survey.
Review of Action Items and Agenda Items for December Meeting	**Scott Strome, Chair, TN RDAC**	