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 July 27th, 2022
8 am CT

RDAC Members in Attendance: Scott Strome, Terry Jo Bichell, Gillian Hooker, Kim Stephens, Chip Chambers, Rizwan Hamid, Lora Underwood, Megan Crow, Richard Finkel,

Others in Attendance: Todd Barber, Ashley McMinn, Skye Miu Steppe, John Fox, Annissa Reed

	Notes	*Action Items*
Call to order and Welcome **Welcome to New Member, Dr. Richard Finkel**	**Scott Strome, RDAC Chair** Introduction of Dr. Richard Finkel from St Jude. Dr. Finkel is a pediatric neurologist working in translational neuroscience, has focused on neurogenetic diseases - e.g. muscular dystrophies and inherited neuropathies. Has participated in clinical trials, including those on antisense oligonucleotides for SMA.	
Rare Disease Story - Skye Miu Steppe	**Skye Miu Steppe, FD Warriors** Born with a rare disease called Fibrous Dysplasia - one of the oldest rare diseases in the world. No treatment, no cure - the only thing that helps is preventive surgery At puberty was when the first symptoms started, around 8 years old. It started with headaches, the only thing that would help was sleep. Her mom started taking her to doctors, telling them something wasn't right. Finally, they called Vanderbilt and got her in to see Dr. Kelly. He did a biopsy of her eye brow, and in March 2007, she had her first facial reconstructive surgery. That held through high school. In 2015, had a second surgery, right before 1st pregnancy. When he was 2 months old, had to have a 3rds surgery in 2016. 4th surgery in 2018. In 2020, the growth shifted to block her nose and led to a 5th surgery. She lives with a lot of fear of future surgeries. Has had 5 facial reconstructive surgeries to date, has minimal bone left on the side of her face. Has had a significant impact on quality of life, migraines daily, lots of pain. Her goal is to avoid surgery if she can. Her one doctor is at Vanderbilt -	

	Started advocating when she was 20 years old - out of pocket costs for medicine are still really high, we're not protecting patients with state insurance support. She was told that this condition would get better once she grew up, but has subsequently learned that that isn't the case. Hormones lead to flare ups of the disorder. Also struggles to maintain balance between her medical needs and taking care of her family. She lives 4.5 hours from Vanderbilt. She does have a local primary care doc, who will do CTs there and send it to Dr. Kelly at Vanderbilt. Diagnosed at 10, one of her biggest fears was losing her hair. Growing up was a struggle, she was bullied, after her diagnosis, she wasn't allowed to participate in sports. At the same time, there some who don't recognize her condition and the parts of it that are invisible, She has been told she has been lying about her condition. After high school was when she decided to advocate. S. Strome called out both the diagnostic odyssey and the physician access issues associated with rare diseases in the state. What can we do when issues arise around access to the small number of experts for conditions that are intervention intensive.	
Proposal for Change in State Exome Coverage Policies	**John Fox, Illumina** **Audrey Ozols, Illumina** Early Periodic Screening Diagnosis and treatment. Federal law that requires that any state accepting medicaid money provide standard of care screening coverage. Law states that if a pediatrician discovers a developmental disability, then they must pay for diagnostic testing. They believe that genetic testing should be a required benefit under EPSDT program. Worth noting that all of the medicaid programs in the state cover whole exome sequencing, but with recent publication of ACMG guidelines that promote exome and genome as a first line test, that establishes a standard of care. R. Hamid sees the problem as not enough providers ordering these tests, also lots of road blocks to getting coverage Dr. Fox proposes a two pronged approach. 1) approach TennCare about WGS coverage policy 2) Support providers in the state with a standard appeals process for denied claims . Comment from Dr. Finkel - argument is often that doing a whole genome isn't going to affect the treatment- this could help arm them with an additional argument. 2nd - colleagues are often nervous to order a test when they dont' know how to interpret results. Is it a provider problem or an insurance problem.	Consider follow up strategy with TennCare CMO

	J.Fox - EPSDT is an argument against the treatment arguments from health plans. Could the RDAC lobby state to remove the requirement for a change in medical management from the policies for coverage of exome and/or genome? They are proposing that RDAC send letter to TennCare to request WGS coverage and/or meet with CMO of TennCare and provide TN providers with a sample appeal letter. C. Chambers - suggested we generate some extrapolated budget data if we end the diagnostic odysseys for a number of kids in TN.	
TN Expanded Genomic Diagnostics Program update / TN Rare and Undiagnosed Disease Network ***Update on Pilot Project Proposal***	**Rizwan Hamid, RDAC Member** **Scott Strome, RDAC Chair** **Chip Chambers, RDAC Member** **Terry Jo Bichell, RDAC Vice-chair** R. Hamid provided an update on conversations with SimulConsult. Thinking through the board, and the process. Hoping to have clarity in the next week or two. Creating a RedCap survey for physician to communicate. Should finalize the details in the next month or so. Believes it could be implemented in a few months. Considering risk management. Also still considering sequencing plan.	G. Hooker and C. Chambers to meet to discuss state coverage policies for Genome and Exome. Will also consider update for TennCare on the program.
Update from NORD meeting - other RDAC activities	**Chip Chambers, RDAC Member** Joined larger group of RDACs for a collective meeting, shared information from our survey. Some discussion of coming up with a more national survey. Arthur Beisang at UMINN	
RDAC Members Finishing Terms / Succession Planning	**Terry Jo Bichell, RDAC Vice chair** Move this discussion to next meeting - TJ Bichell will circulate updates by email.	
Brief Updates **Pharmacy Advisory Committee Update**	**Reginald French, RDAC Member** Lora sent Reginald the PAC agenda - they will be discussing oncology medications and sickle cell in up coming meeting.	

DUR Update **Department of Intellectual and Developmental Disabilities** **Newborn Screening Program**	**Lora Underwood, RDAC Member** No big updates- they discussed an edit and the support act to look at patients on antipsychotics and opiods. No major updates for this committee. **Terry Jo Bichell, RDAC Vice Chair** Meeting is next week. **Rizwan Hamid** Will circulate by email.	
Review of Action Items and Agenda Items for December Meeting	**Scott Strome, RDAC Chair**	
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