

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.

RDAC Members In Attendance TerryJo Bichell, Kim Stephens, Chip Chambers, Richard Finkel, Rizwan Hamid, Lora Underwood

Others in Attendance: Tracey Lovett, Terri Finkel, Todd Barber, Ashley McMinn

<i>Agenda Item</i>	<i>Notes</i>	<i>Action Items</i>
<p>Call to order and Welcome</p> <p>Welcome New Member, Tracy Lovett</p>	<p>Terry Jo Bichell, RDAC Vice-Chair</p>	
<p>Rare Disease Story</p>	<p>Eben Cathey, Live Like Lou Foundation</p> <p>Shared a personal story of his family living with ALS. His wife is living with ALS 4 years post diagnosis.</p> <p>In addition to living with the challenges, they have been very involved in advocacy work.</p> <p>ALS is an underfunded disease - affects 1/300 people, but at any one time ~ 20,000 people are living with ALS.</p> <p>Referenced the Ice Bucket challenge, and the large amount of money raised.</p> <p>One problem with the way advocacy is currently set up - so much is focused on maintenance of the disease and finding ways to live with the disease.</p> <p>And it is difficult to manage.</p> <p>Live Like Lou started in PA, includes some service focused activities, but also very much on research. Partnered with UPenn - to create matching program to build an ALS research laboratory together to find treatments and/or cures. They are doing very basic science. They see this as the current bottleneck - finding the basic scientists who need funding, who need pathways to NIH grants.</p> <p>Looking at the success in PA, E. Cathay's family decided to do the same thing at Vanderbilt - got a commitment from vanderbilt for 1.75 M - and LLL would raise the other \$1.75M - they met this goal in 2 years. Lots of tireless fundraising, awareness, events, getting an audience with the governor's office. They asked for an appropriation directly</p>	

	<p>from the governor and he allocated \$1M. Now hiring a director to manage this research center- Now looking for next opportunity to grow further to \$5M. Director soon to be announced.</p> <p>Questions from RDAC</p> <p>Chip Chambers - part of Rare as One initiative under Chan Zuckerberg foundation- 50 different organizations. He doesn't know of any organization that has raised that much money from the state or federal government. What helped this move forward? Working with governors office, timing was important, handful of advocates in the state house and congress. ALS is a rare disease, but not as rare as other conditions. Looking to other states as example Support from vanderbilt.</p> <p>Needs and coverage for people with ALS - many shift to medicare for coverage. Needs intensify over the course of the disease. Some treatments that cost as much as \$15K / year - apply to foundations for coverage - the HealthWell foundation is one.</p> <p>Big picture -if you're a relentless advocate for yourself, you can find a way to get the things your family needs. But that doesn't feel fair.</p> <p>Even physicians who treat ALS are for the most part standard of care - high focus on how you're worse than the last time you're here - they feel like they don't need to hear that.</p> <p>They have traveled for clinical trials 35 times to Boston or Chicago - 2X they have looked fro the most promising clinical trial - and made a choice- knowing that they only had 3 years to get on a clinical trial - it's a tight window to find the right trial. One involves stem cell injections into your spine. Now on the Healy platform trial - modeled after a clinical trial for cancer, drugs go in - if things go well, you move forward. Innovative way to get drugs to a lot of people. For ALS it felt like a big victory to have this trial become available.</p> <p>Other families in TN living with ALS - in contact with a lot of people battling the disease in one form or another. Communicate usually via email - they led an effort to find families who could contact their legislators - Rep. Cooper is a member of the ALS caucus & on Facebook - finding other people in the state who can act as advocates. Usually it's facebook thats the easiest way to find people.</p> <p>There's a map of people in TN - you can see everyone who has joined their advocacy org - 100's of people in TN - people in Memphis, Knoxville, Nashville tend to have an easier time plugging in.</p> <p>Other folks have to drive a long way to get somewhere that makes sense for them - may have to drive hours for care. Most small hospitals don't have specialists.</p> <p>How are families learning about clinical trials - most hospitals will only educate you about the trials at their own hospitals. People are learning a lot from the advocacy orgs. The platform trials are new.</p>	
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<p>BeginNGS</p>	<p>Dr. Terri Finkel Professor and Associate chair of Pediatrics at UTHSC, Vice-chair of faculty affairs at Le Bonheur</p> <p>Partnering together to end the diagnostic and therapeutic odyssey of rare disease</p> <p>How do we do a better job of diagnosing diseases earlier?</p> <p>Partnership between Le Bonehur and Rady Children’s. Drawing on history of blood spot storage and consent at Le Bonheur - 30,000 consented samples. 15K DNA samples, regeneron offered to do the sequencing for free- now a 100K genome project.</p> <p>Effort is made possible by dramatic improvements in cost and time to diagnosis, increase in number of diagnosable genetic disease</p> <p>GTRx - genome to treatment - automated guidance for clinical management of genetic disorders</p> <p>Delphi model to decide on the list of disorders to include in the screening program</p> <p>Two manuscripts from Rady group have been publsiehd</p> <p>Finding many kids with effective treatments in this group - 57%</p>	
<p>RDAC Planning</p> <ul style="list-style-type: none"> - Meeting Schedule <i>Vote to move to bimonthly meetings</i> - Open Roles - Officer Roles 	<ul style="list-style-type: none"> - Terry Jo Bichell - Gillian Hooker <p>RDAC Members voted to move the meeting to a bimonthly schedule.</p> <p>There are open positions on the RDAC.</p>	<p>Gillian will follow up with Todd to schedule next meeting (November 2022, January 2022)</p> <p>Please send any suggestions for new appointees to Terry Jo Bichell and Gillian Hooker.</p>
<p>TN Rare/Undiagnosed Disease Workgroup</p>	<p>Ashley McMinn Rizwan Hamid Terry Jo Bichell Gillian Hooker Chip Chambers</p> <p>Group has been meeting, will have a meeting later today.</p> <p>Have had some follow up conversations with Simulconsult - seeking to get a proposal from them.</p> <p>Dr. Brown also spoke on a recent White House panel.</p>	

Brief Updates Pharmacy Advisory Committee Update DUR Update Department of Intellectual and Developmental Disabilities State Newborn Screening	<p>Introduced new RDAC nominee - Tracy Lovett.</p> <p>Reginald French, RDAC Member</p> <p>Tracey Lovett, RDAC Member</p> <p>Terry Jo Bichell, RDAC Vice Chair</p> <p>Rizwan Hamid R. Hamid has been giving updates to state newborn screening on the activities of the RDAC. State Newborn Screening is considering RUSP alignment -</p>	
Review of Action Items and Agenda Items for December Meeting	<p>Scott Strome, RDAC Chair</p>	