

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.

Agenda January 26th, 2022

Members In Attendance: Scott Strome, Terry Jo Bichell, Gillian Hooker, Kim Stephens, Chip Chambers, Megan Crow, Reginald French, Lora Underwood

Others in attendance: Kristen Hatcher, Ashley McMinn, Todd Barber

<i>Agenda Item</i>	<i>Notes</i>	<i>Action Items</i>
Call to order and Welcome	Scott Strome, RDAC Chair	
Patient Story	<p>Linda Pilkinton, Bright Heart Foundation</p> <p>Shared the story of her son's diagnosis of hypoplastic left heart, the hope given to them by their cardiac surgeon. They have 2 sons, 1 older and one who has CHD. Her son has had 3 surgeries, 1 at 7 days, 1 at 3 months, and 1 at 4 years.</p> <p>EOB says \$3.5M/ year in care.</p> <p>Cited limited advocacy and awareness in this area.</p> <p>They need awareness, funding, financial support, emotional support, therapies</p> <p>She started BrightHeart Foundation - themes of "Survive, Thrive and Alive"</p> <ul style="list-style-type: none"> - in the hospital, their kids are fighting to survive, especially in infancy. - Thrive is about PTO, OT, helping find the tools that help them get past the diagnosis - Alive - feeling alive, addressing the mental health side of these diagnoses - both the parents and the kids, the adults who are affected, the health care providers <p>Q: Kim Stephens @ organization - when do they get involved? A: Its tricky- there's a break in the situation - the healthcare system is taking care of sick kids and doesn't have the bandwidth to provide the ancillary support. They catch many of them on social media. But that's a hard time - Also when people are in the fog of diagnosis, it's hard to communicate about resources. They started a TN CHD coalition to connect folks together to reduce redundancies. Then she went to AHA and asked them for help - they sent information about heart attack, stroke, etc - she communicated back to them that that</p>	

	<p>was not helpful- She is now petitioning the AHA and conversations are progressing now</p> <p>Q: @ advocacy for research vs. support for patients - She advocates for both, but found more grant money for programs and some of it was a pragmatic decision to go after the most accessible projects, tools, etc to enrich lives today.</p> <p>Her vision for the future: to avoid transplant, to have a cure, to have more awareness.</p> <p>One potential cure is in utero fetal surgery - could you make a heart grow with stem cells? Could you grow a new heart? Could you find interventions that have fewer lifelong side-effects?</p>	
<p>TN Expanded Genomic Diagnostics Program update / TN Rare and Undiagnosed Disease Network Update</p>	<ul style="list-style-type: none"> - Rizwan Hamid, RDAC Member - Scott Strome, RDAC Chair - Chip Chambers, RDAC Member - Terry Jo Bichell, RDAC Vice-chair <p>Updates: Exploring all of the ways to provide faster diagnoses to patients and access to expert across the state.</p> <p>Having project management in place is providing important structure.</p> <p>Making progress on discussing logistics, areas of focus, how to set up phases, thinking through pragmatic approaches and leveraging the multi-disciplinary nature of the group advancing this initiative.</p> <p>Also making progress with follow up w/ Dr. Kingsmore - though there are also many complexities to think through there.</p>	<p>Consider review of state insurance policies for coverage of exome and/or genome</p> <p>Group will continue to meet and move toward a proposal for the project.</p>
<p>2022 Planning In person meetings? New Appointments New Initiatives TennCare needs</p>	<p>Scott Strome, RDAC Chair</p> <p>The group may consider an in person meeting at a later date in the year, pending the pandemic- It was noted that there is not funding for the RDAC, so we would need to consider to that as well. Would likely need to in Nashville</p> <p>Names of reappointees were submitted to governor's office : Scott Strome Lora Underwood</p> <p>Also submitted Dr. Richard Finkel as a new appointee to represent St. Jude to replace Suzanne Jackowski</p> <p>We are awaiting an answer</p> <p>Several people will be coming up this summer: John Callison Chip Chambers Gillian Hooker</p>	<p>All who are coming up on term limits this year should let Terry Jo know if they are able to continue</p>

	<p>Megan Crow Kim Stephens</p> <p>Thinking about areas where we have gaps on the committee - would it be useful to have a pharma perspective. We could look into whether we should have a representative from LifeScienceTN. For the state PAC, industry comes, and there is a designated time period for public testimonies. With DUR, there isn't a space for them.</p> <p>Current initiatives include the Expanded Diagnosis program, state survey and monitoring of issues in the state.</p> <p>Right now, we have 88 for the survey - moving forward</p> <p>K. Stephens also proposed RUSP alignment - federal RUSP is a suggestion to the states of what to put on newborn screening. MPSII is on the docket to be added to federal RUSP. Every Life foundation works on this legislation</p> <p>How do we increase our visibility in the state - start to catalog the rare disease organizations - build relationships that will increase our visibility. Consider working with students who might want a project - C. Chambers also mentioned a project to coordinate with medical students.</p>	<p>Consider speakers from the therapeutic world to come in and give the way they approach it - share thoughts on that.</p> <p>Ask R. Hamid to communicate whether TN is considering adopting the federal RUSP list.</p>
<p>Brief Updates</p> <p>Pharmacy Advisory Committee Update</p> <p>DUR Update</p> <p>Newborn Screening / Genetics Advisory Committee Updates</p> <p>Department of Intellectual and Developmental Disabilities</p> <p>Rare Disease Virtual Hill Day Upcoming</p> <p>https://everylifefoundation.org/rare-advocates/rare-disease-week/</p>	<p>Reginald French, RDAC Member Lora Underwood, RDAC Member <i>Upcoming Meeting February 10th. The agenda will be circulated to the RDAC if we are able.</i></p> <p>Megan Crow, RDAC Member -Notes circulated with agenda</p> <p>Discussed Federal RUSP & need to follow up w/ R. Hamid</p> <p>This group met in November - discussed a number of initiatives. T. Bichell will circulate notes following the meeting.</p> <p>Rare Disease Day is next month, Feb 28th - lots of events around the state - that might be an opportunity to bring attention to the council through our February meeting.</p> <p>Also consider hosting a state-wide rare disease symposium.</p>	<p>All members should consider registering for the the Rare Disease Hill Week</p>