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 April 27, 2022

RDAC Members In Attendance: Scott Strome, Terry Jo Bichell, Kim Stephens, Rizwan Hamid, Megan Crow
Others in Attendance; Todd Barber, Ashley McMinn, Rose Gallagher, Danae Bartke (HCU Network America), Abby Trotter (LifeScienceTN)

Call to order and Welcome	**Scott Strome, RDAC Chair**	
Newborn Screening for Homocystinuria in Tennessee	**Danae Bartke, HCU Network America** Executive Director, HCU network Adult living with HCU Today focusing on classical homocystinuria, as identified by newborn screening Genetic disorder on the RUSP, tested for in all 50 states. When it is missed, elevation fo metabolite - homocysteine - leads to blood clots, strokes, lens dislocation. When she and her brother were born, the state of IL did not have newborn screening. They weren't diagnosed until her brother was ~ 5 - he had missed nearly every milestone An ophthalmologist recognized that he likely had a genetic condition, referred him to genetics where he was diagnosed and the rest of the family was also tested. She was the only other siblight to be diagnosed. When treated from birth, outcomes are a lot better when treated with a low protein diet. She did develop a blood clot in college and has had her lenses replaced. Her family's story is not uncommon There are still a lot of patients missed (est 25-50%, could be as high as 80%) HCU network america founded in 2016 - state and federal policy, also involved in the medical nutrition equity act - we have a mandate for NBS, but no mandate for required treatment. Working on advancing the dx and treatment for HCU and related disorders. They fund research through grants	**Action item:"** **Develop some language about how we liaise with the state genetics advisory committee on issues related to newborn screening. Add a link to the newborn screening website on our websites.** **Revisit follow up on HCU in 3-6 months to see if conversations have progressed with the Genetics Advisory Committee.**

	Strive to improve identification and outcomes of patients Rizwan - TN uses methionine as the analyte and there is a push toward total homocysteine or a ratio based screen with total homocysteine as as second tier. In Qatar - states use total homocysteine as a screening tool and pick up is much higher. There is a worry about methionine taking too long to elevate at birth. Q about process of adding a new analyte to panel? Patient support groups come forward to advocate - chairs of the advisory committee will take it over and do the background research - process can take up to a couple of years Q - what's the best way for our RDAC to make a recommendation to Genetics Advisory Committee? We now have a subcommittee of the RDAC that will liaise with that committee. Meetings are public, we will make sure we are sending representatives - quarterly meetings - is there a place on our website or on Genetics Advisory Committee website to give people who would like to advocate at the state level for these issues - how would we make this a process? Most GACs welcome this. We could provide additional support for the GAC when they are advising addition of a new disorder or changing the metrics - there is a well-established path for this. Q @ good models for medical formulas - in Illinois - formulas are provided free by the state to kids with metabolic disorders. Thre are states that do have programs, often times income based, don't tackle into consideration other expenses - Her formula, out of pocket, would be over $25K per year, another medication is $1800 per month. They are lobbying for medical nutrition equity act at the federal level - that would end a lot of the loopholes - Sarah Chamberlain - PKU news is leading much of that policy effort.	
TN Expanded Genomic Diagnostics Program update / TN Rare and Undiagnosed Disease Network *Update*	**Rizwan Hamid, RDAC Member** **Scott Strome, RDAC Chair** **Chip Chambers, RDAC Member** **Terry Jo Bichell, RDAC Vice-chair** Meeting later this afternoon The challenge - we have a document for circulation - different people at the meetings all of the time. The goal is to get all parties together and circulate comments. Everyone has had a chance to review that draft. Today - make some final decisions regarding next steps. Discussion will be more robust after we've had this meeting this afternoon - then a longer time to share R. Hamid- making good progress. Building relationships, e.g. with illumina. Key next step, once materials are finalized, to meet with Tenncare to make sure they are in the loop on this effort.	**Group will meet on 4/27 to discuss**

Newborn Screening Opportunities ***Update***	**Kim Stephens, RDAC Member** **Rizwan Hamid, RDAC Member** **Abby Trotter, LifeScienceTN** Group asked to present to the genetics advisory committee at meeting tomorrow (4/28/22) Abby, Kim and Erin Frey will present to GAC about oour RDAC. They will provide some information about the council and what it does. Map of the different states and where they are in the NBS RUSP status - in TN, we are one fo the better states in the country- cover all of the newborn screening BUT we do not have a state law to codify that any screening that comes down from the RUSP will be added to the NBS. Tennessee is leading the nation in number of genes screened for on NBS Q @ newborn hearing - is that part of the RUSP? Looking into this. Vision screening is not normally a part of NBS, though is often done in the hospital. In Texas there is a new bill being considered to create an NBS preservation account. In Kansas, NBS for every baby at no charge, also creates a fund to support treatment and referral systems for patients that screen positive. Looking at the landscape, they are thinking about opportunities in TN to improve our newborn screening process. Q - asked about administration of these preservation funds - thought is that they might be funded out of the medicaid programs. There is some concern about the privately funded models of newborn screening being a mechanism to pull funding from newborn screening. There may be opportunities to streamline the paper work process for individuals after they turn 18 - Q - Is there a medicare option here?	**Will report back at next meeting in follow up to this meeting**
RDAC Step Therapy Working Group ***Update***	**Reginald French, RDAC Member** **Megan Crow, RDAC Member** **Kim Stephens, RDAC Member** Step therapy has passed the house and the senate with final language and is on its way to the governor's desk - this bill wil regulate step therapy and create an exception process that is more streamlined and easier. They took tenncare out of the bill, so it was less controversial - it's thought that the governor will sign this next week. It will apply to private insurance only.	

Brief Updates **Term Limits / Succession** **Pharmacy Advisory Committee Update** **DUR Update** **Newborn Screening / Genetics Advisory Committee Updates** **Department of Intellectual and Developmental Disabilities**	**Terry Jo Bichell, RDAC Members** *Waiting to hear from the governor's office on confirmation of Dr. Richard Finkel.* *Alert the UT government relations staff so that they can take that to the governor's office.* **Reginald French, RDAC Member** **Lora Underwood, RDAC Member** *Update provided by email.* **Megan Crow, RDAC Member** *Updated provided by email.* **Rizwan Hamid, RDAC Member** *Meeting tomorrow, update on next call* **Terry Jo Bichell, RDAC Vice Chair** *Meeting in two weeks, will have update on next call*	
Review of Action Items and Agenda Items for December Meeting	**Scott Strome, RDAC Chair**	**Have Tanya schedule a call w/ Rizawan, Chester, Scott, Chip, Abby to discuss collaboration with TennCare.**