Tennessee Rare Disease Advisory Committee (RDAC)

Minutes

RDAC Members in attendance: Scott Strome, Terry Jo Bichell, Rizwan Hamid, Richard Finkel, Chip Chambers, Tracey Lovett

Others in attendance: Kerri Engebrecht, NORD, Todd Barber, UTHSC, Jim Henderson, Acadia, Alex Tabraue, Resta Pharmaceuticals, Clare Judkins, Acadia, Holly Murphy, Acadia, Robert Jaramillo, Reata Pharmaceuticals, Roya Mostafavi, Le Bonheur, John Fox, Illumina, Ashley McMinn, Vanderbilt, Jaimie Patrick, International Rett Syndrome Foundation, Mike Mattoon, Acadia, Kemi Olabasi, Acadia, Abby Trotter, LifeScienceTN, Carley Riehle, Takeda

Agenda Item	*Notes*	*Action Items*
Call to order and Welcome	Scott Strome, RDAC Chair	
Update on TN Diagnostics Network Project	Ashley McMinn, Project Manager Rizwan Hamid, RDAC Member Background to the TN Rare Disease Board Goal for today- take a vote to see if full committee agrees to move forward with this plan and proceed with implementation PlanL Create a network for of rare disease specialists and general physicians to provide guidance to pediatricians caring for kids with rare diseases Overview of project PCP refers case for review by TNRDB - diagnosis identified through case review OR not identified - Patient encouraged to undergo either Project FIND-OUT or subspecialty evaluation Project would start with a 6 month trial to see what kind of outreach comes in then consider approaching the state legislature with a more formal proposal	Project will reach out to the TNGCA to let them know about this project

	As they build the network, could also include genetic counselors S. Strome - Upcoming meeting of deans across the state - theme of the meeting - what can we do together to improve the health of the state. Consider adding a 30 minute window to the agenda to present to the group R. Finkel - Was the committee formally charged with a task, are we addressing that formally with this? Have we considered metrics to measure success. S.Strome - Statorily, we are asked weigh in on drugs in tn, we also want to bring new technologies to TN. We are also allowed to do other things that will benefit the health of Tennesseans in TN - and this falls into that broader category. Question about genomes/exomes - RDAC Vote to support the project: All in favor: Scott Strome, Terry Jo Bichell, Rizwan Hamid, Richard Finkel, Chip Chambers, Tracey Lovett Next steps: We need to meet again, standardize the metrics, still need to reach back out to Dr. Wu to let him know we are working on things, and need to develop the REDCap/decide on outreach. I haven't heard anything from Dan Harder so I am going to need Dr. Strome's help to reconnect with him (his team was going to do the REDCap development for us).	
Rett syndrome & the first and only FDA-approved treatment for Rett syndrome	Kemi Olabisi PharmD, BCPS Holly Murphy Acadia Pharmaceuticals Inc Jaimie Patrick International Rett Syndrome Foundation TN Family Empowerment Team Background presented on Rett Syndrome MECP2 mutation causes it, but not necessary or sufficient for diagnosis. There is a significant disease burden Jaimie Patrick, Rett Syndrome Foundation - Introduced her daughter to the RDAC, was developing normally, then lost the ability to walk, to babble, to feed herself. Was first	

	diagnosed in Iowa then returned to TN and sought care at Vanderbilt- Saw Dr. Carey Fu at Vanderbilt and finally got a diagnosis. They dove right into therapies. She was really challenged at school and it was suggested she just come home. They enrolled in the Trofinitride drug trial - she turned her entire experience around and she got a sunshine award at school for her attitude. Then in 3rd grade, they asked about TCAP testing - she took 10 days to do it - lots of tears and she finished it. In 4th grade, she finished it in 4 days. Trofinitide have been huge for her quality of life. Q: about cost of drug. They had access to the drug on the trial, it has not yet been approved for payment yet since the drug was approved. There is a bridge program that is offered if you were a part of the drug trial - her daughter is in the bridge program until insurance begins approving it. Medicine is oral or g-tube delivered. Closing comment - drug was approved March 10th for kids ages 2 and up. Most common adverse events were diarrhea and vomiting. Cost of the drug - dose dependent, dosed based weight - will share information for committee to review Also highlight -there's an international Rett syndrome foundation meeting in Nashville in 2 weeks.	
Updates on Skyclarys for Friedrich's Ataxia	Robert Jaramillo Alex Tabraue Reata Pharmaceuticals Skycalrys - for FA- just received FDA approve in February FA - chronic loss of neurological function. Most common form of recessively inherited ataxia Most common cause of death is cardiomyopathy Condition caused by GAA expansion mutations in Frataxin - important for the mitochondria	

	Skyclarys - is an activator of the NRF2 pathway - binds to KEAP1 to promote cell response to stress Clinical trials - improvement in MFARS scores vs. worsening in the comparison group Oral administration, few adverse events Challenge currently is to have payers cover this test.	
RDAC Succession Planning	Terry Jo Bichell, RDAC Vice-Chair	Send email to Scott with positions we have to fill and he will circulate.
Patient Story	Diamond Shriner, Parent to Quinnlee Mae Shriner	Will reschedule for the July Call
Brief Updates to circulated by email Pharmacy Advisory Committee Update (see circulated notes) DUR Update Department of Intellectual and Developmental Disabilities Genetic Advisory Committee / Newborn screening	Reginald French, RDAC Member Tracey Lovett, RDAC Member - PAC meeting was cancelled due to not having a quorum in May - moving on to the next quarter - August 10th is slated. - Drugs come in on PA, and then get criteria after reviewed by PAC to determine what criteria will be applied - Discussion about how the RDAC can better support conversations about how to better deliver new and emerging rare disease therapies in the state. - DUR meeting - planned - July 25th Terry Jo Bichell, RDAC Vice Chair Rizwan Hamid- Will be moving forward with MPSII on the NBS in TN	T. Lovett will pass along agenda as it becomes available
Review of Action Items and Agenda	Scott Strome, RDAC Chair	Next meeting July 26th

Items for December Meeting		Action items to consider - rare disease drug in development by Strom e