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 November 24th, 2021

RDAC Members In Attendance: Scott Strome, TerryJo Bichell, Gillian Hooker, Chip Chambers, Megan Crow, Lora Underwood, Reginald French, Kim Stephens, Suzanne Jackowski

Others in attendance: Jessica Duis, Ashley McMinn, Cole Green, Stephen Kingsmore, Todd Barber

Call to order and Welcome	**Scott Strome, RDAC Chair** **Recently joined a panel with other RDAC leaders via NORD** **Thinking about how to support other RDACs and the lessons learned**	
The Angelman Emergency System	**Jessica Duis, MD, Children's Hospital of Colorado** Presented on work from their Center of excellence for individuals with chromosome 15 disorders, including Angelman syndrome **Community Needs** In clinical trials for new therapy for Angelman, lots of travel to centers of excellence, it requires access to experienced providers It raises equity concerns about who gets access to these treatment trials, and the development of better standards Meeting regularly with KOLs, discussing cases, they could continue to tweak the guidelines and practices - similar to an Echo program. Discussed Echo programs - e.g. for hepatitis C - education to providers to educate and empower them to take care of patients with diabetes and hep C. Rare disease is starting to create these types of programs, often led by patient advocacy July 2021- started a hotline - 2 calls/month - it does have to be the provider who calls, not the family - b/c it's hard to give medical advice directly to families Record data from them, follow up with provider and families using standardized program for advice Biggest challenges- providers have been reluctant to talk,b/c of disconnects between families and providers about how things are going. Showed an example clinical pathway from the children's website - working to make them clickable so that they can track data on the clinical pathways	

	Staffed by a nurse during business hours - family signs a HIPAA form, trying to get the provider on call in touch with the provider who's calling - triage system during the day. If the call is from an ER or PICU, they take the calls immediately. They are getting more calls from international community than national community. And they have interpreter services available. This could work pre diagnosis or post. In the US it's often trainees who are calling. General pediatricians more than neurologists. **Funding** Funding for a full time nurse to take calls and collect data. Implementing quarterly meetings with KOLS to go over all of the cases that have come in. Where might this need tweaking, where might the data suggest that this has been successful vs. not. Lots of details to provide guidance to providers Traditional route has been through patient advocacy routes, but this gets more providers involved. Subcontracts to different providers. There are CPTs for provider-provider consults, but they are not currently reimbursed. They have talked to managed care specialists about this - they need to meet with insurance companies and managed care specialists and medical directors to talk about paths to reimbursement. Ultimate goal is to be able to bill for this. **Research goals** Also making clinical pathways live on the website, launch web-based consultation services - where the provider is located, what their question was, understand what information people want. **Expanding Services** Working to expand into ST/OT, nutrition, education advocate **Questions from the RDAC** TJ Bichell cited the calls that come into her org, and the benefits of having this as a resource for her organization to refer people to refer their doctors to this center. This is a way that treatment could be expanded for all kinds of rare diseases. S.Strome we are working on a model to reduce the length of diagnostic odysseys in TN, given thought to expanding beyond Angelman - how do you justify costs at 2 calls/ month? How do you scale to other disorders? Dr. Duis- They are thinking a lot about this. Right now it's funded by the Angelman community, they thought it would be a lot busier - Presenting to groups that bring together groups of parent advocacy groups to promote a cost sharing model. Thinking about using the same infrastructure for vascular malformations - now have 10 other rare disease groups that have reached out.	
Progress in Pediatric Diagnosis	**Dr. Stephen Kingsmore, Rady Children's** Newborn screening for genetic diseases with effective treatments using whole genome sequencing	

	Currently - using NBS w/ Mass spec- wonderful for the conditions affected- now looking for a screen for roughly 500 conditions . They would like to add to screening panel - genetic disorders for which you need a genome sequence to diagnose Gap- effective therapies becoming available - growing need to bridge the gap Since 2010 - working w/ critically ill newborns - w/ diseases of unknown etiology. There's a rush to make a dx, if wrong diagnosis is made ~ $10K to do this in a trio - gradually becoming standard of care across hospitals. Spotty implementation of this in the US. They do testing for over 70 centers of N. America. Newborn screening - in screening mode- only looking for Reasons why now: Cost of genome is decreasing, time to decode a genome is decreasing, explosion of therapies. E.g. Zolgensma - not finding it's way to the kids, want to do better newborn screening to fix that. It's not just about the test - many infrastructural components are required. Looking at rapid diagnostic genome sequencing - gave them the confidence to move forward into newborn screening Working with a large network of organizations- patient advocacy, pharma, manufacturers, technologists (e.g. TileDB & Genomenon), international groups (e.g. GenomeEngland) How did they decide on the panel - which genes- built a virtual management guidance system for frontline pediatricians, similar to Newborn screening ACT sheets. Retweaking so that it is designed for newborn screening - 403 disorders that are well qualified for screening - there are curative or effective therapies for those conditions - surveyed 10K different treatments for these 560 diseases to arrive at the 403 cases. Technologically, they've taken the WGS test and messing with it to become a screen Wetlab is almost identical to WGS, test menu is quite different - instead of interpreting genetic variants, they look up known variants. Deal with many variants that map to 403 gene-disease dyads. Moving forward, surveying drug candidate and catalogs - goal to is have more disorders with effective therapies. So that they can accelerate the time to add genes to panel - aim for 5 min to add this. This should not replace NBS, but rather be in addition to, synergistic with. BUt there is great national infrastructure for dealing with screen positives - why reinvent the wheel, start with a dried bloodspot - and build into current infrastructure. Right now - at prototype stage - 3 pilots that are going well - they are starting to talk to groups like us about this to think about moving into pilots in kids. Next Spring would like to test 1000 newborns prospectively at a few centers Then would like to move into the powered pilot stage to test 10's of thousands of newborns. 5 year time frame, let this start to deploy nationally	

	Education will be a key piece of succes. Next steps - they want a rare disease community steering group, developing materials for public launch, nominate additional conditions for inclusion, help in public launch of project, assist in fundraising for pilot infrastructure and testing It will be controversial - how to overcome sensitivities of this becoming a policy - messaging will be important. **Questions** Kim stephens - offered to help w/ committee - cited slow process of the RUSP, feels that this is just so needed. Geneticists asked whether it's ethical to talk to parents about clinical trials - she feels it's unethical not to talk about clinical trials when the child has a progressive disease. Upside of WGS approach-cost to add geens goes down significantly. The RUSP aims for perfection, but w a genome, we will have low sensitivity initially - we may only pick up 50% of babies initially. Scott Strome - screening people in collaboration with Regeneron - 35-50 kids/week - sees the challenges in informed consent given potential life-long impact of sequencing, diseases for which we don't have treatments - Start with a rifle shot, work in areas where there are no ethical reasons not to return this information - E.g. every condition looks like SMA type 1 - as long as you do that - not diverging from the RUSP. With time, let society address other questions - Dr. Kingsmore is most worried about false positives - that is the largest obstacle. TJ Bichell- thinking about how we pilot - in TN 80K / births per year - if there were a few sites, what would funding look like - how would we show that it's cost effective- Scott is more worried about ethics than infrastructure. Dr. Kingsmore - their test is $8.5K - 10K, they demonstrate that it delivers QALY gains at an acceptable price.	
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** Ashley McMinn presented on the straw man document presented, coming into next meeting with feedback.	**All of working group -** read through the document and provide comments and feedback.
Brief Updates **Term Limits / Succession** **Pharmacy Advisory Committee Update**	**Terry Jo Bichell, RDAC Members** **Reginald French, RDAC Member** **Lora Underwood, RDAC Member** **Megan Crow, RDAC Member** **Rizwan Hamid, RDAC Member** **Terry Jo Bichell, RDAC Vice Chair**	**Waiting to hear from new contact at St. Jude -** then will send the list to governor's office

DUR Update **Newborn Screening / Genetics Advisory Committee Updates** **Department of Intellectual and Developmental Disabilities** **TN Rare Disease Survey**	**Kim Stephens, RDAC Member** No major updates. M. Crow asked about the American Rescue Plan Act - and whether the RDAC might play a role in influencing decisions- to see where funding was going and whether there is a role for input.	**M. Crow** will follow up with Lora Underwood
Review of Action Items and Agenda Items for December Meeting	**Scott Strome, RDAC Chair**	