

## Tennessee Rare Disease Advisory Committee (RDAC)

Minutes 3/22/23

RDAC Members In Attendance: Gillian Hooker, RDAC Secretary, Reginald French, Rizewan Hamid, Terry Jo Bichell, RDAV Vice Chair, Tracy Lovett

Others in Attendance: Yeyberth Lopez, Christine Escamillia (Reata) , Holly Murphy (Acadia), AShley McMinn (Vanderbilt), Kerry Engebrecht (RAN), Kemi Olabisi (acadia), Roya Mostafavi (Le Bonheur), Abby Trotter (LifeScienceTN), Gene McCarty (Reata)

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| Call to order and Welcome       | Terry Jo Bichell, RDAC Vice-Chair   |  |
| Patient Story - Gaucher Disease | <p>Yeyberth Lopez</p> <p>Yeyberth Lopez, born in Venezuela, Diagnosed at 5- kept getting sick, nosebleeds, swelling. Went to the emergency room, and was diagnosed by a specialist there.</p> <p>At the time of diagnosis, there was no treatment.</p> <p>In 1997-moved to the US b/c St. Jude had reached out to them b/c they thought it was cancer. When they learned it wasn't cancer, they were referred to Le Bonheur</p> <p>1999 - started enzyme treatment from Genzyme</p> <p>After treatment - bone pain was much better, not as severe as it used to be, fewer symptoms.</p> <p>She was told she would never be able to have children - but she married her in the US - had a son.</p> |  |

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|  | <p>As a military wife - limited specialist access - and very few people who knew her disease.</p> <p>Was referred to MUSC for delivery of her son, and she had a doctor from TN who also traveled with her.</p> <p>Her son was tested at birth and does not have Gaucher.</p> <p>Today - if she were to have a baby - she would want to know at birth about Gaucher - She had bone crises, pain, splenectomy,</p> <p>With earlier diagnosis, earlier treatment</p> <p>For her - as a teenager, had to miss a lot of school for treatment. Pain was a factor in not getting to participate in daily life</p> <p>Coverage for therapies is tricky - avoids changing jobs b/c of changes insurance, changing prescriptions</p> <p>Sometimes she has to pay out of pocket - there are programs with assistance - needs to be more organized - maybe plans that specialize in rare diseases</p> <p>Any metabolic genetics clinic will take on Gauchert - Knoxville, Vanderbilt, Memphis - Hematology</p> <p>Enzyme replacement, and oral therapy modifies the lysosomal activity</p> <p>Coverage is a huge problem - lots of pre-authorization letters.</p> <p>Some plans have no coverage for enzyme therapies or genetic genetic therapies - for gaucher it's been around for a while</p> <p>Would be useful to have legislation about coverage for genetic conditions - she also sees her doctors spending a lot of time - would be nice if there was one place to go -</p> |  |
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|  | <p>Patient services - get a card top pay for doctors and medicine.</p> <p>R. Hamid- things insurance companies are eager to drop coverage as soon as they can b/c treatment is expensive.</p> <p>Costs are not dropping, they remain very expensive. Also may have limited facilities.</p>   |  |
| Update on TN Diagnostics Network Project | <p>Ashley McMinn, Project Manager<br/>Rizwan Hamid, RDAC Member</p> <ul style="list-style-type: none"> <li>- Talking to the IT team to design the workflow to allow the patient access to the 2nd opinion aspect of the project</li> <li>- Once that's ready - presentation to this committee about</li> <li>- Website to be launched once the committee gets approval</li> <li>- Narrowing scope</li> <li>- Sending follow up to Dr. Wu</li> <li>- TJ Bichell - presented Project FIND-OUT at ACMG</li> <li>- Thinks we should consider Project FINDOUT as an RDAC project</li> <li>- Project FINDOUT is thinking about going to the Dept. of Health as a pilot project - it's a research study to understand whether this algorithm</li> <li>- The RDAC could endorse this project and also build their diagnostic process as complimentary to FIND-OUT</li> </ul> | <p>-Working group meeting next week</p> <p>-Send follow up to Dr. Wu</p> |
| Freidrich's Ataxia Therapy               | <p>Christine Escamilla, PhD<br/>Gene McCarty<br/>Reata Pharmaceuticals</p>   | Reschedule for future meeting  |

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| RDAC Succession Planning   | Terry Jo Bichell, RDAC Vice-Chair   |  |
| <p>Brief Updates</p> <p>Pharmacy Advisory Committee Update (see circulated notes)</p> <p>DUR Update</p> <p>Department of Intellectual and Developmental Disabilities</p> | <p>Reginald French, RDAC Member</p> <p>Tracey Lovett, RDAC Member</p> <p>New Drug approvals in February</p> <p>Next Meeting - May 11</p> <p>Currently just a skeleton on the website</p> <p>Hutchinson-Guilford Progeria drug on the agenda</p> <p>DUR - April 25th</p> <p>Question about an adrenal insufficiency drug shortage - worries about drug hoarding, etc.</p> <p>Terry Jo Bichell, RDAC Vice Chair</p> <p>They had a great method for making recommendations to state</p> <p>Future meetings - would be good to present on method of making recommendations</p> <p>Would we consider inviting a state senator or congress person on our committee</p> <p>At a certain point in the year, every member of the council makes recommendations, council votes on priorities - they compile and those are sent directly to the director of the department of intellectual and developmental disabilities.</p> <p>Then they report on the recommendations</p> <p>Progress on the recommendations</p> | <p>On the agenda for future discussion</p> <p>- how the RDAC makes recommendations to state and where the RDAC sits in the state</p> |
| Review of Action Items and Agenda  | Scott Strome, RDAC Chair  |  |

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| Items for Next Meeting |  |  |
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