

GENETICS AND NEWBORN SCREENING ADVISORY COUNCIL MEETING

The Genetics and Newborn Screening Advisory Council (GNSAC) meeting was held Friday, February 23, 2018, at the Florida Department of Health Bureau of Public Laboratories, 1217 North Pearl Street, Jacksonville, Florida.

Call to Order:

The meeting was called to order at 9:04 am EST by Paul Pitel, MD, Council Chairperson. Roll was taken and introductions were made.

Members Present:

Paul Pitel, MD, Chairman, Hematologist
Robert Fifer, PhD, Audiologist
Dorothy Shulman, MD, University of South Florida (USF)
Roberto Zori, MD, University of Florida (UF)
Jeffrey P. Brosco, MD, Children's Medical Services (CMS) Deputy Secretary
Carina Blackmore, MS Vet Med, PhD, Department of Health, Delegate for the Surgeon General
Heather Smith, Parent Consumer
George Fox, Parent Consumer
Brian Kirk, Newborn Screening/March of Dimes Representative
Barry Byrne, MD, Practicing Pediatrician

Guests:

Gerold Schiebler, MD, CMS Consultant
Jeanne Brunger, PerkinElmer, Inc.
Larry Vroegindewey, PerkinElmer, Inc.
Christina Vracar, AHCA
Corinn Rich, PerkinElmer
Angela Darnell, PerkinElmer
Sharon Bowden, Pediatrix Medical Group
Susan Weinger, Pediatrix Medical Group
Debbie Hayes, Sanofi Genzyme
Ann Lucas, Sanofi Genzyme
Helen Travers, Sanofi Genzyme
Donna O'Steen (USF)
Amarillis Sanchez-Valle (USF)
Mario Estevez (Project Alive)
Mike McBrierty (Biogen)
Scott Shone (RTI)
Mike and Kelly Strenges (Parent Consumers)
Jhoanny Cardenas (NORD)
Dan Kilbride (Cambrooke Therapeutics)
Alissa Swota (Baptist Jax)

DOH Personnel Present:

Cassandra G. Pasley, JD, BSN, Tallahassee
Marcy R. Hajdukiewicz, MS, Tallahassee
Dusty Edwards, BSN, RN, Tallahassee
Jessica O. Meyer, MSW, Tallahassee
Bonnie Taffe, PhD, MPH, Bureau of Public Health Laboratories, Jacksonville
Patti Ryland, MT, BS, Bureau of Public Health Laboratories, Jacksonville
Patty Parrish, Bureau of Public Health Laboratories, Jacksonville

Ming Chan, PhD, Bureau of Public Health Laboratories, Jacksonville
Sajani Mamallapalli, Bureau of Public Health Laboratories, Jacksonville
Lori Bair, Bureau of Public Health Laboratories, Jacksonville
Melissa Rodriguez, Bureau of Public Health Laboratories, Jacksonville
Ken McCain, CMS
Aaron Matthews, CMS
Rachel Eastman, Contracts, Tallahassee
Emily Reeves, BSN, RN, Nurse Follow-Up, Tallahassee

Attended Via Conference Call:

Allison Westphal, BSN, RN, Tallahassee
Jennifer Moore, BSN, RN, Tallahassee
Su Meter, BSN, RN, Tallahassee
Sandra Crump, RN, Tallahassee
Gary Kleiner, MD, PhD, Practicing Pediatrician
Lori Gephart, APD Representative
Emily Jones, March of Dimes

Dr. Paul Pitel introduced Dr. Barry Byrne as one of the practicing pediatrician representatives for the Council as well as an expert on Pompe Disease.

CMS Division Update

Cassandra Pasley provided the Council a Division of Children's Medical Services update.

There are three fiscal issues related to CMS. Senator Lauren Book presented a member's proposal for \$1,900,000 for Pompe Disease. More to come on this proposal.

The Poison Control Center Information Network, receives \$5,000,000 in general revenue, \$3,000,000 of which is non-recurring. A request has been made for these funds to be recurring.

For the Early Steps Program, starting January 1st of this year, an at-risk category for infants was created, such as infants with Neonatal Abstinence Syndrome (NAS). Additional funds were requested using a previous federal grant to handle the new children that would be added to the program.

There are two substantive items that are being followed. There is a request from the pediatricians in the Child Protection Community to capitalize the references to their program in statute because individuals sometimes interpret it as a service as opposed to a full program.

There is another bill that primarily impacts AHCA regarding infants with Neonatal Abstinence Syndrome (NAS). This bill suggests that infants born with NAS be able to be served at PPECC (Prescribed Pediatric Extended Care Centers) and would make the infants with NAS automatically eligible to be served in PPECC. Also, the Department of Health would be required to contract with a university to analyze the effectiveness of that and how much funds would be saved to see those children in the PPECC as opposed to a hospital setting.

Dusty Edwards updated the Council regarding the process for adding new disorders. On July 1, 2017, statutory language was changed that outlines the timeline for adding disorders to Florida's screening panel that have been added to the RUSP. DOH has one year from the date a disorder is added to the RUSP for the disorder to be considered on Florida's screening panel. Once the Council votes to recommend adding it to the Florida's panel, the state has 18 months to begin screening for that disorder.

Newborn Screening Laboratory Update

Dr. Taffe provided an update to the Council. Dr. Taffe was named Chief Chemist and Laboratory Director. Two Chemist IIIs and two Chemist IIIs have been hired. There is one remaining vacancy, Biological Administrator II, which should be filled in March 2018.

The Quality Assurance turn-around time is an aggregate number of abnormal and the normal. From birth to release is less than 8.29 days overall. The national goal is to report out all specimens within 5 days. Abnormals are reported out sooner, due to priority. Some delays are due to method of shipping, i.e., regular mail vs. overnight shipping. The hurricanes this season significantly affected shipping times. Normally the Laboratory is at 95 to 98% on reporting out in less than 5 days.

Dr. Zori has asked to see the national benchmark versus the benchmark for the Program at next meeting. Dr. Blackmore points out that Florida's birth to report time may be higher than some states due to the lack of a contracted courier service in our state. Dr. Pitel suggests that data be presented at the next meeting to allow the Council to make educated recommendations.

Software Upgrade

A data system upgrade, with PerkinElmer, has been completed to allow for X-Linked ALD testing. An electronic ordering system via web portal has been developed for hospitals to use for specimen card submission. Tallahassee Memorial Healthcare is using the portal now.

DOH is attempting to get Florida's data in a format that can be used the Clear Risk Assessment Tool. This is replacing the R4S assessment tool.

In preparation for X-ALD, the laboratory remodel has been completed to bring in 4 new mass-specs. X-ALD go-live is scheduled for May 1st. The FDA kit is not yet approved. DOH plans to switch the FDA approved kit once it is approved. A 3-tier testing method will be used. The first 2 tiers will be completed by the Laboratory. Genetic sequencing will be completed by PerkinElmer Genomics. Based on numbers from New York (currently screening for X-ALD), Florida should expect to see approximately 40-50 referrals for x-linked ALD annually.

Dr. Taffe provided an update regarding CDC cooperative agreement/grant to fund NextGen sequencing for SCID/CF, and lysosomal storage disorders (LSDs) project with PerkinElmer and UF.

The Laboratory has evaluated the Baebie's Seeker Platform for LSD testing, which is FDA approved. The differences between the two testing methodologies were discussed.

Newborn Screening Follow-Up Program Update

Ms. Edwards provided an update to the Council. The Follow-Up Program received an FTE for X-ALD implementation. An OPS nurse currently with the program received the FTE position and the open OPS position was filled. The data analyst position is the only vacancy, at this time.

The NBS statistics were reviewed with the Council. Dr. Brosco requested clarification regarding open cases from 2015. The Council discussed appropriate length of time for cases to remain undiagnosed.

The program is looking to utilize NewSTEPS case definitions in order to create consistency from a national level. A Memorandum of Agreement with NewSTEPS is being developed in order to submit Florida data into the national data repository.

The web portal is operational at Tallahassee Memorial Healthcare. The Laboratory is looking to hire a project manager to fully assist in this process.

Medical Food and Formula

One of the things needed to do is bring in a project manager that could look at the data and analyze how many people are receiving formula, look at the age limit of those receiving formula, streamlining the process to ensure these families are obtaining the formula. In fiscal year 2016-17, DOH had about 267 individuals obtain formula through the pharmacy. Please note that this is the number of individuals, not number of times an individual went to the pharmacy for formula. The age range was from zero to 71. Of the 267 people, 130 were under the age of 18; approximately half of those receiving formula were children. Examining the data assisted in determining improvements. As a result, looking at the formulary will be helpful. The Genetic Centers in the state have been contacted to send in information based on what is being prescribing to patients. Is this being prescribed because that is what is in the formulary or what is best for the patient?

Cassandra Pasley: DOH decided this needed immediate action and the following has been completed:

- 1) Three genetic centers were contracted for \$25,000 each for a food pantry for individuals to obtain needed food or formula.
- 2) Working with Medicaid to determine why individuals are unable to obtain formula.
- 3) Preparing to give \$25,000 additional dollars to the genetic centers.
- 4) DOH has safety-net dollars that is for any child in need that cannot obtain PKU food/formula.

The Program will focus on PKU at this time.

Dr. Zori stated PKU is the obvious choice. The treatment of PKU has two parts and both use the word food. One is low-protein foods, which is a cost, and the other is formulary, which is a greater cost.

(Discussion regarding the formulary continues)

Dr. Schiebler stated the issue is that the \$25,000 is non-recurring. Legislation has been introduced this year regarding the issue. The issues are:

- How many individuals are there in the state who need this?
- Food is needed for all age groups.
- Insurance companies have a very high deductible.
- Insurance companies have a limit.

The question is: Is this an important enough issue for the Department to introduce legislation next year that will address all components of the issue?

Dr. Brosco suggests that the discussion be tabled until more information is available.

Ms. Pasley suggested hiring a nurse to be responsible for working with PKU. PKU would be the first priority, however, the nurse would also be responsible for assessing all disorders currently screened, one at a time.

Update on X-Linked ALD

Ms. Reeves provided an X-ALD update: Discussed the testing algorithm, follow-up protocols, data system updates, referral process, education for Genetic Centers, and reporting.

Discussion continued regarding about what is needed in the insurance realm for treating these patients as stem cell transplants would be part of the treatment for any with a finding of X-ALD.

Extensive discussion ensued on how to do it well, the adult aspect, and what that would mean.

Newborn Hearing Screening Follow-Up Program Update

Mrs. Jessica Meyer provided the Newborn Hearing Screening Program update. Information regarding staffing and grant updates was provided.

The Newborn Hearing Screening Program has formed an Advisory Committee for EHDI. Appointment letters have been sent out and date of first meeting is pending.

At the Council's request, a parent refusal survey was conducted by the Hearing Program and the results were discussed.

Dr. Pitel requested additional information be provided at the next meeting regarding the 1-3-6 benchmarks.

Approval of Minutes from previous meeting

Dr. Pitel asked for the minutes to be accepted. Dr. Shulman so motioned and George Fox seconded her motion. Minutes were accepted at 11:23 a.m.

Discussion on Framework

The Follow-Up Program worked with RTI International to create standard criteria for the Council to use when considering new disorders, develop a framework using the developed criteria, and use the developed framework to provide information for Pompe Disease and MPS I that is state specific.

The Council discussed the benefits of using a framework when considering the addition of new disorders to Florida's screening panel.

Dr. Alissa Swota

Dr. Swota is a medical ethicist from Baptist Jacksonville and addressed the Council regarding medical ethics when considering the addition of new disorders to Florida's screening panel.

Extensive discussion continued on the ethics of when to inform, testing, and outcomes.

Vote to add MPS I and Pompe

Dr. Pitel lead the voting discussion:

1. Once the Council votes yes, there is an 18-month clock that starts, where the State has to implement.

2. Council members agree that this vote should move forward.
3. Council members agree there is a great deal of complexity to begin screening these disorders.

The question for the Council is: Do we have a way to move forward with the complexity of doing it and making it work?

Extensive discussion ensued on the pros and cons of waiting with a request for additional information from the states that currently screen and for Dr. Taffe to expand her testing.

Public Comment

Mario Estevez gave his family's story. Mr. Estevez works with Project Alive and his son was diagnosed with Hunter Syndrome.

Ms. Pasley recommended that a committee, or sub-group, be formed with Dr. Byrne and Mr. Fox to create a plan for submission at the next meeting.

An additional request was made for RTI to provide additional data. Ms. Pasley informed the council that the job was complete at the submission of the report. Anything in addition to that would have to be put out for bid or solicitation.

Committee votes on Pompe – unanimous no –

Committee votes on MPS I – unanimous no -- Voting for both disorders took place at approximately 2:08 pm EST.

Dr. Schiebler would like to know more specific information regarding pulse oximetry, i.e., who is screening, etc. and have this data available at the next meeting. Dr. Schiebler believes it should be in statute as it would then be required.