

GENETICS AND NEWBORN SCREENING ADVISORY COUNCIL MEETING

The Genetics and Newborn Screening Advisory Council meeting was held on Friday, February 19, 2016 at the Florida Department of Health Bureau of Laboratories, 1217 N. Pearl St., Jacksonville, Florida.

Call to Order:

The meeting was called to order at 10:05 am EST by Paul Pitel, MD, Council Chairperson. Roll was taken and introductions were made.

Members Present:

Paul Pitel, MD, Chairman, Jacksonville
Carina Blackmore, MS Vet Med, PhD, Tallahassee
David Auerbach, MD, Orlando
Dorothy Shulman, MD, Tampa (USF)
Heather Smith, Consumer, Lakeland
Keith Nash, March of Dimes, Orlando
Lori Gephart, RN, Agency for Persons with Disabilities, Tallahassee (via teleconference)
Mark Weatherly, MD, Orlando
Robert Fifer, PhD, Miami (UM)
Roberto Zori, MD, Gainesville (UF)

Guests:

Barry Byrne, MD, PhD, University of Florida
Brittany James, Audiology Extern
Callie Ellis, Pediatrix Medical Group
Christian Gentile, Audiology Extern
Elisa Seeger, Aidan Jack Seeger Foundation
Jeanne Brunger, PerkinElmer, Inc.
Kelly Strenges, ALD Parent
Larry Vroegindewey, PerkinElmer, Inc.
Mike Strenges, ALD Parent
Peter Grisgby, ALD Parent
Sharon Bowden, Pediatrix Medical Group
Sierra Lawrence, Audiology Extern
Susan Weinger, Pediatrix Medical Group

DOH Personnel Present:

Bonita Taffe, PhD, MPH, Bureau of Public Health Laboratories, Jacksonville
Cassandra Pasley, BSN, JD, CMS, Tallahassee
Dusty Edwards, RN, BSN, CMS, Tallahassee
Lois Taylor, RN, BSN, CMS, Tallahassee
Ming Chan, PhD, Bureau of Laboratories, Jacksonville
Penelope Edwards, MS, RD, LD, CMS, Tallahassee
Patty Parrish, Bureau of Laboratories, Jacksonville
Patti Ryland, Bureau of Laboratories, Jacksonville

Susan Meter, RN, CMS, Tallahassee, (via teleconference)
Whitney G. Jones, CMS, Tallahassee

2016 Legislative Update

Lois Taylor gave the council a legislative update. Two bills were introduced to the legislature that affected the Newborn Screening Program (NBS) which would add Adrenoleukodystrophy (ALD) to Florida NBS Panel. They have stalled as of now, but may be reviewed at a later time. ALD and MPS1 were added to Recommended Uniform Screening Panel (RUSP) on February 16, 2016.

Dr. Pitel reiterated that the process for adding a disorder to the Florida NBS panel is a medical decision not political decision. The council will evaluate each disorder as they are added to the RUSP.

Ms. Taylor announced her impending retirement to the council. The advisory council awarded Ms. Taylor with a plaque recognizing her exemplary service to the Newborn Screening Program and to the families of Florida.

Guest Speaker: Mike Strenge and Elisa Seeger

Mike Strenge, a parent of a child with ALD, gave the advisory council a presentation about ALD.

Elisa Seeger, founder of The Aidan Jack Seeger Foundation and a parent of a child with ALD, gave the advisory council a presentation about ALD.

Representative Mike La Rosa offered his support in implementing ALD on the Florida newborn screening panel and in exploring funding for adding this disorder.

The council discussed adding ALD to the Florida Panel and the ability of the Florida state lab to implement ALD testing, issues of funding for the implementation of screening, and concerns related to payment of providers for treatment with the current reimbursement available through Florida Medicaid. Dr. Taffe noted that there is not an FDA approved kit available for ALD screening at this time, however one may be approved in 2017.

Dr. Robert Fifer made a motion to add ALD to the Florida newborn screening panel. The motion was seconded by Dr. David Auerbach and Dr. Dorothy Shulman. The motion passed unanimously. The Department of Health will begin researching cost and impact in adding ALD to the Florida panel.

Guest Speaker: Barry Byrne, MD, PhD

Dr. Barry Byrne gave the advisory council an update on Pompe and the status of the IRB proposal. The approval from Dr. Peter Iafate and the Institutional Review Board (IRB) at

University of Florida has been completed. PerkinElmer has proposed that they will assume all costs during the provisional period as a way to validate their assay for Pompe screening.

Dr. Byrne also spoke to the council about Duchenne muscular dystrophy (DMD) and the current therapy before the FDA for approval.

Newborn Screening Laboratory Update

Dr. Bonnie Taffe gave the advisory council an update on the Newborn Screening Laboratory. The lab has three new chemists recently hired. Proficiency testing was passed in the third and fourth quarter. Also, turnaround times for the lab shows a slightly lower turnaround for abnormal results.

Go-live is planned for end of March for the new SCID assay. The current assay kit for Cystic Fibrosis has been discontinued. A new assay has been evaluated and go-live is at the end of February.

PerkinElmer is developing a neo lysosomal storage disorders kit with an application before the FDA. PerkinElmer has proposed an IRB to work with the lab to conduct a retrospective study to document the sensitivity of the new test.

The retention of blood spots was discussed among the council regarding the costs, space, and validity of blood spots after periods of time.

Newborn Screening Follow-Up Program Update

Lois Taylor gave the advisory council an update on staffing changes within the Newborn Screening Follow-Up Program. There are currently four vacant positions within the follow up program all in various stages of being filled. Dusty Edwards is the new program director over the follow-up unit. Also, Penny Edwards has been hired to analyze data for the program.

Dusty Edwards gave the advisory council an update on the Newborn Screening Follow-Up Program. APHL Newborn Screening and Genetic Testing Symposium is being held in St. Louis, Missouri. It is being attending by Bonne Taffe, Ming Chan, Cassandra Pasley, and Dusty Edwards.

Dusty Edwards reviewed the program statistics handout that was provided to the council and all in attendance.

The program is currently working with DOH legal to update the Florida Administrative Rule Chapter 64C-7: Prenatal and Postnatal Risk Screening and Infant Screening for Metabolic, Heredity, and Congenital Disorders.

The Laboratory began scanning specimen cards in January 2016. This increases the Follow-up Nurses ability to obtain information that may be on the specimen card in a timely manner and eliminates laboratory resources used to pull the original card.

Electronic Laboratory Ordering/Electronic Laboratory Reporting (ELO/ELR) first Go-live date has been postponed until March for Baycare Hospitals.

Dusty Edwards gave the council an update on the Newborn Screening Hearing Follow-Up Program. The Hearing Program was awarded a continuation of both CDC (Centers for Disease Control and Prevention) and HRSA (Health Resources Services Administration) grants. The focus of these grants is to maintain and enhance the data system with regard to hearing screening and to reduce lost to follow-up after not passing the newborn hearing screen.

eReports™ now has 288 registered users who are entering the majority of hearing screening data that was previously faxed and being entered by CMS staff.

A hearing screening statistics handout was provided to the advisory council.

New Discussion Items

Dr. Roberto Zori complimented the lab on their dedication to serving the people of Florida.

Dr. Zori spoke about the issue of Medicaid rejecting paying for genetic testing and the issues surrounding that.

Dr. Dorothy Shulman asked if the Newborn Screening Program could provide data for the TSH and T4 screens since testing has been done for 5 years. Shulman offered to review the data and provide input. The Department agreed.

Heather Smith noted that the workgroup for blood spot storage as proposed in the previous advisory council meeting was not yet created, but will be created at a later time. The advisory council unanimously approved the July 31, 2015 minutes.

Adjournment

The meeting adjourned at 1:47 pm EST.