

## **GENETICS AND NEWBORN SCREENING ADVISORY COUNCIL MEETING**

The Genetics and Newborn Screening Advisory Council meeting was held on Friday, January 25<sup>th</sup>, 2013 at the Florida Department of Health, 1217 Pearl Street, Jacksonville, FL.

### **Call to Order:**

The meeting was called to order at 10:02 a.m. EST by Paul Pitel, MD, Chairman of the Council. Roll was taken and introductions were made.

### **Members Present:**

Paul Pitel, MD, Chairman, Jacksonville  
Mary Beth Vickers, RN, MSN, Tallahassee (CMS)  
Robert Fifer, PhD, Miami (UM)  
Dorothy Shulman, MD, St. Petersburg (USF)  
Olaf Bodamer, MD, PhD, FACMG, FAAP, Miami (UM)  
Bonnie Hudak, MD, Jacksonville  
Roberto Zori, MD, University of Florida (UF)  
Heather Smith, Orlando  
Lori Gephart, RN, Tallahassee  
David Auerbach, MD, Orlando  
Susanne Crowe, BPHL, Jacksonville

### **Guests:**

Linda Carter, PerkinElmer, Inc.  
Joseph Levin, PerkinElmer, Inc.  
Elen McCune, Gainesville, University of Florida (UF)  
Charlie Williams, MD, University of Florida (UF)  
George Fox  
Sharon Bowden, St. Vincents

### **DOH Personnel Present:**

Lois Taylor, RN, CMS, Tallahassee  
Erin Obernier, CMS, Jacksonville  
Laura Olson, CMS, Tallahassee  
Donna Barber, RN, CMS, Tallahassee  
Drew Richardson, CMS, Tallahassee  
Pam Tempson, MS, CMS, Tallahassee  
Jasmin Torres, BPHL, Jacksonville  
Jojo Dy, MD, BPHL, Jacksonville  
Ming Chan, MD, BPHL, Jacksonville  
Patti Parrish, BPHL, Jacksonville

## **Conference call:**

Cyril Blavo, MD, Nova (Council Member)  
Marna Zok, RN, CMS, Tallahassee  
Allison Westphal, RN, CMS, Tallahassee

## **Housekeeping/Reminders**

Dr. Pitel reminded the council members to turn in their lunch money, return their travel vouchers, to review the minutes from the previous advisory council meeting, and to turn off all cell phones and beepers.

## **2013 Legislative and Staffing Update – Mary Beth Vickers**

Ms. Vickers provided an update on the leadership positions at the Department of Health and Children's Medical Services.

Department of Health Leadership positions:

- Dr. John Armstrong – DOH State Surgeon General
  - Mead Grigg – DOH Deputy Secretary for County Health Departments
  - (Vacant formerly Kristina Wiggins) – Deputy Secretary of Administrative Functions
  - Dr. Dennis Cookro – Deputy Secretary for Health and Children's Medical Services (CMS)

Children's Medical Services Leadership

- Lynn Marie Firehammer – Bureau Chief of Early Steps retired
- Tobi Goodman – Unit Director - retired

Michael Cantens is the Department of Health Legislative Planning Director

Mrs. Vickers made a motion to reelect Paul Pitel as Chairman of the Advisory Council. The motion was seconded by Dr. Zori. Unanimous vote to reelect Dr. Paul Pitel as Chairman.

## **Legislative**

Committee meetings have been ongoing since December 2012.

There are two bills that would impact the Newborn Screening Program, and they are identical.

SB124 – Senator Jeremy Ring – Adding CCHD to Florida's Newborn Screening Panel  
HB81 – Representative Cary Pigman – Adding CCHD to Florida's Newborn Screening Panel

## **Dr. Hudak's Cystic Fibrosis Mutation Panel Presentation**

Dr. Zori discussed standardization of genetic counseling and asked if we could get genetic counselors licensed in Florida. Dr. Hudak responded that counseling for cystic fibrosis (CF) is very different.

Collect information on how counseling is handled with CF in Florida.

Action item: Report back to the Genetics and Newborn Screening Advisory Council Meeting with comparison of the CF Foundation recommendations to what is being done in Florida.

The CF mutation panel presentation compared Florida's panel against national registries to try to determine if Florida is using the most appropriate CF DNA mutation panel.

- Data from 2007 – 2011 reviewed.
- 80% of CF mutations identified were on Florida's panel.
- CF Foundation wants IRT cutoff below 60. Florida is 45 – 50 (better than what is asked).
- .5% highest incidence mutation that wasn't on Florida's panel.
- Florida's panel contains 86% of alleles identified in over 400 people born with CF in Florida.
- 2.6% of patients missed based on Florida's panel.
- Next steps – compare IRT distribution vs. sweat test results.

Conclusion: Estimates are consistent and are within the range considered acceptable by the Cystic Fibrosis Foundation (CFF).

### **Severe Combined Immuno Deficiency (SCID) Testing Update**

On November 29<sup>th</sup>, 2012 DOH held a press conference to announce testing for SCID.

Testing began on October 1<sup>st</sup>, 2012. The first patient was identified with possible SCID on October 25<sup>th</sup>, 2012. Less than 20,000 specimens were tested when the first SCID patient was identified. Confirmatory testing showed patient as having SCID. The type of SCID was not one of the 17 identified SCID variants.

Another patient had 0 TRECs, but died before confirmatory testing could be performed.

No additional patients have been identified since screening began.

### **Laboratory Update – Jasmin Torres**

Considering adding total galactose (TGal) as second tier screening for Galactosemia (GALT).

- One reagent set is enough for 80 samples.
- One reagent must be used within 60 minutes after reconstitution.
- New kit available in Summer Of 2013 with stability of 7 days for 4 plates.
- Not enough repeat test to run TGal, daily; might be possible with new kit.
- TGal as second tier test can potentially reduce referral for GALT. (New reporting algorithm needed)

The number of specimens received by the lab continues to decrease year over year.

Subcommittee on GALT testing – Dr. Zori is the lead and will report back at the July meeting.

### **Newborn Screening Update – Lois Taylor**

A year ago it was reported that the Newborn Screening Follow-up program had staffing issues due to losing the University of Florida staffing contract. Newborn Screening and all of the Department of Health have suffered staffing turnovers. There are efforts underway to recruit and retain employees. In December of 2012 the Follow-up program lost 2 more nurses.

National Newborn Screening and Genetics Resource Center (NNSGRC) – lost the contract with HRSA and was replaced with "NewSTEPS" – national registry for reporting newborn screening information and statistics.

Newborn Screening statistics on rates of abnormal specimens and referrals were provided. There weren't any statistics that generated questions or concerns.

The Follow-up Program is no longer follows the day of birth hypothyroidism cases - Neonatal Hyperthyrotropinemia. These babies get a repeat specimen, and will continue to get the recommendation to get a repeat specimen. This is a reduction in over 1000 cases per year. Dr Shulman approved the change.

The Follow-up Program is trying to reduce borderline workload, so we can focus more on presumptive positives.

Statement that was previously approved: If we have a repeat specimen that is for a term baby (more than 37 weeks of gestational age), there is an abnormal borderline, and baby is on TPN, we don't refer these babies. The statement then lists several disorders that are not referred. We would like to modify this statement that was previously approved by the Genetics and Newborn Screening Advisory Council, to remove Galactosemia as one of the disorders. A Galactosemia borderline is treated the same as a presumptive positive.

Dr. Pitel made the motion to accept changed language. The vote was unanimous in favor of accepting this updated language.

Florida Newborn Screening Results (FNSR) System – Multiple accounts can be registered under each physician. We are adding more data to this system.

Florida's newborn screening disorders are not listed in statute. They are grouped together in the Rule. 2008 was the first year that disorders were specifically listed in the Rule and it is anticipated that the rule will be amended to add SCID.

### **Critical Congenital Heart Disease (CCHD) Presentation – Dr. Auerbach**

January of 2012 the Genetics and Newborn Screening Advisory Council endorsed CCHD screening.

CCHD is screened using pulse oximetry.

The original CCHD bills from 2012 (HB 829 and SB1052) stated that non birthing centers must refer babies to a physician for CCHD screening within 10 days. These bills were different. They set the date of implementation as October 1<sup>st</sup>, 2012. Florida Medical Association (FMA) and the CMS Cardiac Subcommittee supported the program. These bills did not pass during 2012 session.

CMS Cardiac subcommittee tasked members to report on the status of pulse oximetry screening in their institutions.

A lot changes in the first 48 hours of life. Is the 10 day timeframe requirement for referral appropriate?

Council would like to see national cost savings data for CCHD screening.

DOH has received direction from Dr. Armstrong – If wearing CMS Cardiac Subcommittee or Genetics and Newborn Screening Advisory “hat” and meeting with a legislator, Michael Cantens must be involved and aware of the meeting.

The GNSAC has already recommended CCHD. The Newborn Screening Program does not need legislation to implement CCHD. It only needs funding appropriations.

Motion Dr. Patel – We reaffirm our support for the program looking for funding to appropriately implement CCHD.

Motion 2<sup>nd</sup> by Dr. Fifer. Vote unanimously in favor of Dr. Patel’s motion.

### **Hearing Screening Update – Pam Tempson**

Mrs. Tempson provided performance data on the hearing screening program. The hearing program’s performance indicators of all babies being screening by 1 month, diagnosed by 3 months, and enrolled in services by 6 months are improving year over year. Incidence rates of hearing loss in the state of Florida (1.5 – 2 per 1000) are within the national averages.

Focus on lost to follow-up.

Florida is considered a leader state for hearing screening.

Council members asked for late onset hearing loss numbers.

Provider rewards program – Hearing screening reviews hearing screening statistics on a monthly basis to provide awards for the top performers. This incentive program was expanded to include Early Steps offices for completing IFSPs, quickly.

Update on recruitment of audiologists statewide to become CMS-certified audiologists. Florida currently has 81, which is 4 more than we had at the previous meeting, and the State of Florida is at an all-time high for CMS approved audiologists.

Update on ongoing Meet Me calls. CEU credits are provided for participants. Physicians will now be eligible for CEUs as well. We will be targeting that audience for the next upcoming Meet Me call.

### **Information Technology Update – Drew Richardson**

Drew Richardson gave the update for the E-Reports. It will allow the audiologists, or hospital staff, to enter hearing screening results directly into a website, allowing direct access more quickly, real-time. Hopefully this will cut down on letters going out to parents that should not go, i.e., asking for parents to have their babies rescreened when they've already been rescreened but the results have not made it to the State office yet.

Another option we are checking into for E-Reports is for a screen to be designated for CCHD.

### **Hearing Update – Dr. Fifer**

Dr. Fifer discussed a work group for a national directory of facilities along the East coast, for audiologists, physicians and parents. Should be live and operational by the end of March. It's goal is for use for ages newborn through of 5.

### **Confirmatory Testing – Dr. Bodamer**

Dr. Bodamer gave an update on his workgroup on how to do confirmatory testing for metabolic disorders in the state of Florida. Case definitions must be in place. Other states were discussed and how they diagnose. Independently of the working group, there are 2 other working groups, one is HRSA and the other is SERGG. Also discussed the use of a confirmatory template, once agreed upon by all referral centers in Florida, to go through the list of conditions. UF, USF and UM are the three referral centers that the Newborn Screening Program works through. Examples of questions to be addressed: should it be done in-hospital, outpatient; what is the cost associated with both; time from screening to diagnosis.

### **Future Discussion**

Discussed moving all Genetics and Newborn Screening Advisory Council Meetings to Jacksonville.

### **Public Comments**

There were no public comments.

### **Minutes approval**

The minutes from July 2012 were approved

### **Adjournment**

The meeting adjourned at 2:25pm EST.