

GENETICS AND NEWBORN SCREENING ADVISORY COUNCIL MEETING

The Genetics and Newborn Screening Advisory Council meeting was held on Friday, January 20th, 2012 at the Florida Department of Health Bureau of Laboratories, 1217 Pearl St., Jacksonville, FL.

Call to Order:

The meeting was called to order at 10:06 a.m. 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)
John Waidner, MD, Jacksonville
Robert Fifer, PhD, Miami (UM)
David Auerbach, MD, Orlando
Dorothy Shulman, MD, St. Petersburg (USF)
Cyril Blavo, DO, Fort Lauderdale (NSU)
Melissa Joiner, Tallahassee, March of Dimes
Olaf Bodamer, MD, PhD, FACMG, FAAP, Miami (UM)
Bonnie Hudak, MD, Jacksonville
Melissa Perez, Tallahassee
Max Salfinger, MD, Tallahassee (BOL)
Penny Edwards, MS, representing Roberto Zori, MD
George Fox, Gainesville
Lori Gephart, RN, Tallahassee (via conference call)

Guests:

Linda Carter, PerkinElmer, Inc.
Joe Levin, PerkinElmer, Inc.
Elena Perez, MD, Tampa
Alitta Boechler, University of Miami
Molly Vangorp, University of Miami

DOH Personnel Present:

Lois Taylor, RN, CMS, Tallahassee
Laura Coleman, CMS, Tallahassee
Dusty Edwards, RN, Tallahassee
Drew Richardson, CMS, Tallahassee
Pam Tempson, MS, CMS, Tallahassee
Jasmin Torres, Bureau of Laboratories, Jacksonville
Jojo Dy, MD, Bureau of Laboratories, Jacksonville
Patty Parrish, Bureau of Laboratories, Jacksonville

Ming Chan, PhD, Bureau of Laboratories, Jacksonville

Conference call:

Donna Barber, RN, CMS Tallahassee
Allison Westphal, RN, CMS Tallahassee
Manouchka Pierre, RN, CMS Tallahassee
Rachel Eastman, CMS Tallahassee
Bill Blanchard, MD, Nemours Pensacola

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.

Introduction:

Dr. Pitel welcomed Mary Beth Vickers, RN, MSN of Children's Medical Services, to the council as the Department of Health representative. He also welcomed Dr. Olaf Bodamer, PhD, FACMG, FAAP, the new University of Miami representative joining the advisory council.

2012 Legislative Update

Ms. Vickers provided an update on the recent legislative activities in regards to the Department of Health and Children's Medical Services. There are two pieces of legislation currently being proposed, HB 1263 and its companion bill, SB 1824, which call for the restructuring of the Department of Health. The legislation first proposes to eliminate the CMS Network Advisory Council, a longstanding council that has been relatively dormant for the past 6 or 7 years. CMS has proposed to retain this council for the following reasons: the council will likely play a prominent role in the future as CMS moves forward with integrated care systems statewide and Medicaid reform; in addition, the Cardiac Subcommittee functions under the umbrella of this council, which is of concern to CMS as well. The legislation also reduces the responsibilities of the Department of Health, three of which will impact CMS: Child Protection Team, Perinatal Services and Early Intervention services. However, it was understood that those services should be covered within the new responsibilities listed under the legislation. The legislation also proposes to fill the CMS Deputy Secretary position, to combine the CMS Network and Related Programs and the Prevention and Intervention Divisions, and to retain the Poison Centers within CMS. The Governor's recommendations included replacing Early Steps funding of approximately \$3.6 million and did not propose to pick up DOH's LBR for \$6.9 million for Early Steps; that is now back on the table and being negotiated. Recommendations also included putting back into place the non-recurring dollars for CPT and Poison Centers.

Dr. Salfinger noted how differently the Bureau of Laboratories will be affected by the reorganization legislation. Under the proposed structure, a new division named the

Division of Emergency Preparedness and Community Support will be created, as well as some additional shifting of other divisions. Dr. Salfinger suggested that the Laboratory commonly works across multiple divisions so these changes would most likely not propose difficulty.

Dr. Fifer asked if the \$6.9 million referred to earlier is the money that is required to qualify for the Federal Management Part C.

Mary Beth Vickers answered that that is correct.

She continued with a discussion of the of the Newborn Screening Follow-Up Program staffing issues. When the staffing contracts with the University of Florida ended, the follow-up staff employed under those contracts were moved to much less desirable contracts. Ms. Vickers and Lois Taylor met with State Surgeon General Dr. Frank Farmer on the mission critical status of the Follow-Up program and how essential those positions are to the program. Also, an emergency meeting of the Genetics and Newborn Screening Advisory Council was called and a letter was generated for submission to Dr. Farmer regarding the issues. Under the current contract, benefits are greatly reduced which has led to staff loss and a lack of recruiting potential. Dr. Farmer agreed to assist in seeking vacant nursing positions in the field and reclassify them into the program. Two nursing positions were identified and are currently being processed for the Newborn Screening Program. In addition, the LBR request for new full time employees (FTEs) was amended to request eight new positions in CMS through the use of vacant County Health Department positions. Ms. Vickers commended the program and Ms. Taylor on the functioning of the program under such duress.

Dr. Bodamer asked about the potential liability issues if the program is running understaffed.

Ms. Taylor replied that there is a risk because of the loss of more experienced staff and knowledge from within the program.

There was additional discussion on the rippling effects and impact that such changes could create within the state. Dr. Pitel offered an example of these impacts: In 2010, Florida Medicaid covered roughly 70% of actual costs to deliver care at Wolfson Children's Hospital; in 2011, Florida Medicaid paid 55%; if the Governor's proposals take effect, that number would reduce to 35%. He stated that the hospital would not be able to function with the same quality and comprehensive system of care under a 35% reimbursement rate. Additionally, some children's hospitals could lose between 1/2 and 2/3 of the pediatric faculty. The majority of Children's Hospitals throughout Florida could likely suffer such effects.

Critical Congenital Heart Defect (CCHD) Screening

Dr. David Auerbach provided a presentation on an overview of Pulse Oximetry Screening for Critical Congenital Heart Disease. Topics discussed included:

- Orders - what we are doing
- Algorithm - how the process flows
- Process and equipment concerns
- Screening Results
- Family feedback

George Fox asked if there was a list of the different conditions that the testing could possibly reveal. Dr. Blanchard (conference call) replied that patients with heart muscle issues would most likely not be revealed from the testing, but rather those with oxygenation issues. The screening is an attempt to locate those babies that are in the well-baby nursery who otherwise appear healthy. Mr. Fox added that a list of potential disorders picked up by this screening could be beneficial to promote the adoption of the testing.

William Marvin, MD, Pediatric Cardiologist at Wolfson's Children's Hospital, stated that approximately 8 out of 1,000 children are born with Congenital Heart Disease. The testing could potentially pick up secondary disorders from other causes.

Dr. Blanchard added that CCHD screening is supported among cardiologists throughout Florida and should be adopted under the screening program. There may be potential issues in training rural hospitals and physicians on screening techniques; however that should not bar the adoption of the screening in Florida.

James Mosteller, the Florida Government Relations Director at the American Heart Association, provided a presentation detailing Pulse Oximetry Screening to detect Congenital Heart Defects.

Penny Edwards of the University of Florida asked about the follow up process for CCHD screening.

Dr. Pitel discussed the follow up process for CCHD. The screening and follow up is most analogous to hearing screening. One question that will arise is the how to deal with hospitals that do not have trained pediatric cardiologists or access to necessary training due to rural locations. Also, an issue that may arise is the Governor avoiding any programs that will cost the state additional money.

Dr. Bodamer suggested that in order to recommend disorders added to the panel arguments must be made to support that addition, such as cost-effectiveness and added benefit to society in terms of saved lives. Also, some hospitals and states already screen for CCHD and it would benefit Florida to look at that data and compare as a tool for education and promoting the screening. A prospective study would be an excellent method of doing this.

Dr. Auerbach stated that a quality improvement approach could benefit as opposed to a research approach.

Dr. Salfinger added that it's important to question if this screening should be a part of Newborn Screening as the majority of the screening actions for CCHD takes place at the hospital, rather than the laboratory as in the testing for other disorders.

To attempt to answer Dr. Salfinger's question, Dr. Pitel asked why hearing screening was added to the newborn screening panel.

Dr. Fifer replied that many programs come down to cost savings.

The group further discussed the cost-savings and developing a data analysis in order to support screening for CCDH.

Melissa Joiner of the Tallahassee March of Dimes asked if a bill analysis was performed on the current legislation to determine cost of implementation.

Lois Taylor responded that a bill analysis was done on CCHD screening. The majority of cost will be incurred by hospitals, specialists, technicians, and equipment. The Newborn Screening Follow Up program will handle referrals and potential loss to follow-up patients. A referral process would need to be established.

Ms. Joiner asked Mr. Mosteller if New Jersey performed a bill analysis, he responded that they did and he could provide that if requested. Ms. Taylor added that CMS utilized the New Jersey bill analysis when producing theirs for Florida.

Dr. Pitel asked the council how it would like to proceed.

Dr. John Waidner responded that it's possible that a cost-effective analysis may be difficult to prove; it would be better to implement the program because it's the right thing to do.

Dr. Pitel asked the council if it would be possible to vote, based on data and national recommendations, in favor of exploring CCHD screening. The council agreed without further discussion. Dr. Pitel asked for further discussion on the next steps to follow in order to make a recommendation to add CCHD to screening.

Dr. Salfinger stated he was still unsure if this testing should be added to the NBS panel.

Dr. Pitel stated that the council approved and would like to make a recommendation, but would like further instruction on implementation and other logistics from the Cardiac Subcommittee and Cardiac specialists. If the council approves, a letter will be drafted and moved forward to the appropriate parties. A motion was made and passed with no dissent.

SCID Update

Dr. Elena Perez, an Immunologist from All Children's Hospital, provided a presentation on Severe Combined Immunodeficiency Disorder (SCID).

- True incidence: ~1 in 46,000 and in some populations, ~1 in 22,000
- 5-10 cases per year in FL
- A total of 17 cases in the last 5 years in FL (some cases missed)
- Upfront costs of SCID to FL
- 20% early diagnosis (>3 months of age), usually due to family history
- Comparison of early vs. late diagnosis - 94% survival rate with early transplant

Dr. Salfinger stated that there would need to be additional staff (FTEs) to perform the testing and follow-up activities. Outsourcing would be possible. Additional budget authority would be needed. The overall laboratory budget would be increased by ~\$2 million.

Dr. Pitel stated that the decision comes down to: save babies' lives at relatively low costs, or lose lives at a very high cost.

Dr. Perez agreed that the choice is between having a worse outcome and greater cost (without SCID testing) or greater outcome and lower cost (with SCID testing).

Dr. Pitel summarized the political background behind the SCID testing issue. Representative Matt Hudson's bill was vetoed at the last moment by the Governor. It was suggested that DOH should move forward with SCID testing on the agenda, however that was retracted as the Governor's office stated that they would not be supporting any bills that they had vetoed last year. More recently, it was suggested that this may again change.

Dr. Perez stated that Senate Bill 550 is in the works. Heather Smith (SCID Angels/parent), on conference call, discussed the recent political environment in relation to SCID. Ms. Smith stated that there is a great deal of miscommunication and misinformation regarding SCID testing. She asked if there would be a fee increase if SCID were added to the testing. Mary Beth Vickers replied that there would not. Lois Taylor added that the fee being spoken of is the \$15 hospital fee per live birth. Although this fee to the hospitals would not increase, there would be an increase to what is being billed to private insurance companies and Medicaid by \$16.67 per specimen. Ms. Smith said that this was the issue in the Governor's office. Dr. Pitel added that the cost benefit to the taxpayers would still be greater if SCID was added.

Ms. Smith and Dr. Perez stated that there needs to be greater education with this issue.

Ms. Vickers declared that she and Ms. Taylor developed a white paper for implementing SCID and provided that information to the legislative planning director who provided it to the Governor's office. They also met with lobbyist Doug Russell to discuss SCID implementation, who is also moving forward with educating the appropriate parties.

There was additional discussion regarding providing education to insurance company lobbyists about the cost savings of SCID testing.

Dr. Pitel discussed how the council would move forward from that point.

Dr. Fifer suggested that the council vote to reaffirm the importance of the issue of implementing SCID screening from a public health perspective as well as a fiscally sound perspective on the statewide level. He made a motion, which passed with no dissent.

Dr. Pitel concluded by discussing the creation of a letter for dissemination. He would draft the letter; Dr. Perez offered to contribute. Ms. Vickers suggested that the letter be consistent with the white paper previously provided, in order to prevent more confusion.

The meeting was suspended at 12:40 for lunch.

Laboratory Update - Jojo Dy, MD

Dr. Dy provided an update on the state laboratory's recent activities and issues. Topics discussed included: quality assurance activities; Dr. Piero Rinaldo's review of MS/MS protocols and suggestions for changes; changes to Congenital Hypothyroidism; automation of instrumentation at the laboratory; and potential future changes.

Dusty Edwards, RN, BSN, provided an update on TSH with the Newborn Screening Follow-Up Program.

Penny Edwards asked if it was possible to get a list of positive predictive values for the list of disorders screened for in newborn screening. Jasmine Torres replied that they would be able to provide that for the council.

Dr. Shulman commended the lab on an excellent job on turnaround times. There was a recent patient with Congenital Adrenal Hyperplasia who was identified quickly and who would have most likely died if not for the quick turnaround time on the lab specimen. Jasmine also discussed a recent baby who was found with low citrulline, which is not looked at under newborn screening, and the baby was referred out with a critical disorder because of the diligence of the laboratory employees. The council further commended the lab on a job well done.

There was a discussion regarding the issue of confirmatory testing and dealing with insurance companies. Dr. Bodamer suggested that the University of Miami could potentially process specimens for diagnostic testing after the laboratory is properly set up and certified; Dr. Pitel requested an update when the process was complete.

Dr. Dy continued his update of laboratory activities.

Hearing Screening Update - Pam Tempson

Pam Tempson, MS, provided an update on hearing screening in Florida. She detailed statistics over the past year related to newborn screening hearing follow-up activities.

Dr. Fifer discussed the progress of the development of a pediatric audiology roster on a state and national level, and issues with loss to follow-up. The rosters will be beneficial for parents and providers to access based on patient needs.

Ms. Tempson continued with an overview on recent changes and updates in the hearing follow-up program.

Newborn Screening technology updates—Drew Richardson

Drew outlined the implementation of Direct Service Messaging email system. The system will enable a method of sending protected health information (PHI) securely and efficiently. E-reports should be starting during the summer, initially with hearing and possibly with other disorders following later.

Newborn Screening Update—Lois Taylor

Lois Taylor, RN, BSN, provided an update on the recent activities of the Newborn Screening Follow-Up Program. In 2011 the birth rate stabilized after decreasing for a number of years. Physician requests totaled 87,000 for 2011; 74,000 of those were obtained through FNSR. The system has been extremely beneficial to the Newborn Screening Program. Phone calls have increased tremendously. Ms. Taylor discussed how the follow-up program handles borderline cases and program statistics in the past year.

It was recommended that Dr. Phil Ferrell review the Cystic Fibrosis program; the program has not been officially reviewed since its implementation in 2007. Ms. Taylor asked the council to make a recommendation that CMS request for Dr. Ferrell to review the Cystic Fibrosis program. Dr. Auerbach made a motion for this action; the motion was seconded and passed with all in favor. The entire program was reviewed in the past (1992 and 2004) by the National Newborn Screening and Genetics Resource Center (NNSGRC) at no cost. Another review of this type would be beneficial to the follow-up program. A motion was passed to request another review of the entire Florida Newborn Screening Follow-Up Program by the NNSGRC. The motion passed with no dissent.

Ms. Taylor reviewed the staffing issue currently afflicting the Newborn Screening Program.

New Business

Melissa Joiner asked if there were any disorders on the panel that did not need to be screened for, because of the proposals to add new ones. Dr. Pitel and Lois Taylor replied that removing disorders from the panel would not save costs and that Florida should continue following the national standards for screening.

Dr. Bodamer suggested that the Newborn Screening Program should provide a update of the children saved through screening, and the positive long-term outcomes screening provides. There is an initiative to create a long-term database to enable this information to be made available. Dr. Bodamer will provide an update to Dr. Pitel at the next meeting.

Dr. Pitel discussed the Minnesota articles provided on blood sample retention policies. The articles detail the Minnesota Supreme Court's opinion on whether the Minnesota Department of Health violated the state's genetic information laws.

Dr. Auerbach stated that there needs to be a dialogue regarding potential risks and benefits for retaining blood specimens. Florida's currently has a 6 month retention policy and does not allow retaining samples for research.

The group discussed the issues related to blood spot retention policies and whether it would be sound to propose an opt-in or opt-out for quality assurance purposes.

Dr. Fifer stated that United Healthcare has begun a program of direct sales to its enrollees in Florida. The program is based on an online hearing test that evaluates two frequencies only, uses uncalibrated equipment, and programs a direct sales hearing aid on the basis of an invalid audiogram. The program may violate license law in Florida and is an effort currently being fought. One concern is that parents might take a hearing test on behalf of their child in order to purchase a lower cost hearing aid without the proper procedures needed to safely and properly operate the hearing aid.

The other issue Dr. Fifer offered to the council was a program that Medicare would be unveiling in the fall called value-based purchasing. Under the program, physicians will be paying a set fee to cover all services and supplies for a specified period of time. The program may force a change in mindset in healthcare practice outside of the hospital setting, and may eventually affect pediatric therapy services.

Melissa Perez updated the group on recent issues of Department of Health and Human Services (HHS) releasing the preliminary list of central benefits without follow-up treatment for inborn errors of metabolism. Interested groups were able to obtain representation at the meetings. The HHS then relegated the decision to decide what is covered under central benefits to each state.

Public Comments

There were no public comments.

Minutes approval

The minutes were approved with a few minor changes.

Adjournment

The meeting adjourned at 3pm EDT.