

GENETICS AND NEWBORN SCREENING ADVISORY COUNCIL MEETING

The Genetics and Newborn Screening Advisory Council meeting was held on Friday, July 13th, 2012 at the Florida Department of Health, 4025 Esplanade Way, Tallahassee, FL.

Call to Order:

The meeting was called to order at 10:04a.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)
Melissa Joiner, Tallahassee, March of Dimes
Olaf Bodamer, MD, PhD, FACMG, FAAP, Miami (UM)
Bonnie Hudak, MD, Jacksonville (via conference call)
Melissa Perez, Tallahassee
Helen McCune, representing Roberto Zori, MD
George Fox, Gainesville (via conference call)
Lori Gephart, RN, Tallahassee

Guests:

Linda Carter, PerkinElmer, Inc.
Kara Bechtold, University of Miami
Emily Steffel, University of Miami
Andrea Pilna
Philicia Adams, Pediatrics
Yorquiz Perez Mendez, Pediatrics
Susan Weinger, Pediatrics
Heather Smith, SCID Angels for life
John Smith, SCID Angels for life
Taylor Dahley, SCID Angels for life
Gul Dadlani, M.D., All Children's Hospital
James Mosteller, American Heart Association

DOH Personnel Present:

Lois Taylor, RN, CMS, Tallahassee
Stefanie Higgins, CMS, Tallahassee
Laura Olson, CMS, Tallahassee
Dusty Edwards, RN, CMS, Tallahassee
Chrishonda Jenkins, RN, CMS, Tallahassee
Drew Richardson, CMS, Tallahassee
Pam Tempson, MS, CMS, Tallahassee
Jasmin Torres, Bureau of Laboratories, Jacksonville
Jojo Dy, MD, Bureau of Laboratories, Jacksonville

Conference call:

Jeff Jacobs, MD, All Children's Hospital
Elena Perez, MD, University of Miami

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.

2012 Legislative Update – Mary Beth Vickers

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 that were discussed at the previous meeting in January, HB 1263 and its companion bill, SB 1824, which call for the restructuring of the Department of Health.

After the January update, Matt Hudson placed a strike all bill to the house version, which was drastically different. This included organizational changes – structural and operational. Children's Medical Services spent next few months meeting with legislative staff and DOH staff to voice concerns. The end result came with many changes within CMS, but the final version much more palatable than it was in the beginning. It was a much better outcome than original bill. The major changes that occurred in the bill are:

1. Addition of term "serious" to the definition for children with special healthcare needs, children enrolled must have chronic and serious healthcare condition.
2. Eliminate authority to provide service pregnant women or eligible children; Must be enrolled.
3. Safety net population (5-6k children) – children that do not qualify for Medicaid or title 21 but costs for health care spends them down in to financial eligibility. A sliding fee schedule will be created for this population.
4. Adoption – historically children who were adopted and received any medical or surgical services prior to adoption would remain in the network. Now these children will need to have a serious and chronic health condition.
5. Consolidation of two divisions – CMS Network and related programs and Division of Prevention and Intervention – child protection team, poison, telemedicine, sexual abuse treatment programs.
 - a. Consolidated two divisions and Relocated programs and staff
 - b. Name is now "Division of Children's medical services"

CMS has been meeting regularly with executive staff at DOH for guidance to implement new provisions. Almost everything is complete with the exception of safety net, as it requires rule promulgation and incorporation of a sliding fee schedule. We have a green light on all other provisions

There were significant changes in staff and structure. There are a reduced number of divisions within the department and bureau.

- a. New State Surgeon General, Dr. Armstrong , came on at the end of May
- b. Made a few more structural changes
 - i. Deputy secretary for health – Steven Harris
 - ii. Deputy secretary for CMS – vacant for 18 months (Dr. Chiaro)

iii. Deputy secretary for administration – Kristina Wiggins

The new revised structure moved CMS from under Kristina Wiggins to Dr. Harris. Dr. Harris' new title is Deputy Secretary for health and Deputy state health officer for CMS

The Deputy Secretary for CMS position will be filled. This position oversees all operations for the county health departments.

The Chief of Staff position, which was filled by Richard Solze, but has been vacant since December/January, will also be filled. This position is responsible for overseeing the office of legislative planning, office of communications and quality management

Ms. Vickers and others from CMS have been meeting every week with Dr. Harris to get him up to speed without overwhelming him.

There were two budget requests submitted during legislative session; the first was to convert the contract staff to FTE's. The second request came directly from Dr. Farmer. We did receive FTE's for the UF contract staff that was affected. Unfortunately, we took a budget cut of \$2.5 million, so we are unable to fund all of the FTE positions. 8 FTE positions were requested and we were able to fill 5. This was a success. We were not able to fill the other positions, but we are much better off. We currently have 3 follow up nurses in FTE's and 2 contract nurses. This will hopefully reduce turnover. We were also able to convert Pam Tempson and Drew Richardson into permanent positions.

Paul Pitel asked the council if a letter should be drafted requesting funding for all FTE positions
Moved by Helen McCune

Paul Pitel will draft a letter to send out to council for approval

Fifer second

All in favor

Ms. Vickers stated that the other victory/accomplishment during session was the implementation of SCID on the NBS panel

Dr. Farrell's Cystic Fibrosis Review – Bonnie Hudak

Dr. Hudak shared that they were tasked with undergoing an audit. Dr. Phil Farrell, from University of Wisconsin, was asked to conduct the audits. Dr. Farrell worked with Donna Barber in the Newborn Screening Program to conduct the audits in March and April 2012. Dr. Farrell and Ms. Barber visited 3 CF centers in Florida. They toured sweat labs, conducted meetings, followed up with conference calls with CF center directors and people actually doing NBS and counseling.

Dr. Farrell reported his findings; Bonnie Summarized the report.

Dr. Farrell complimented the labs for their efficiency and noted the great work by Donna Barber.

99.5% of newborns were screened

1.89% unsatisfactory specimen rate

Incidence of CF 1:6137 (predicted incidence 1:4000). The incidence is lower than predicted; lower incidence rate in Miami Dade and Broward areas. This is most likely due to Hispanic population and lower rate of CF in that population

The goal for CF centers is to notify families within 3 days of referral from NBS and get the referred infant in for sweat test within 1-2 weeks. There are no immediate consequences of delayed diagnosis, but if you look at outcomes at 10 years of age, children diagnosed through NBS do better than children diagnosed by symptoms.

Age at notification of families – outstanding; mean 12 days, median 13 days

Age at diagnosis or case closure – excellent; mean 43 days, median 21 days

QNS rate for sweat tests – tolerably low; 8.2%. CFF guidelines call for <10%

Dr. Farrell's recommendations included:

Review mutation panel for CF. Over 1800 mutations have been demonstrated, but not all of these are disease causing mutations. The panel we use tests for about 40 mutations. This was collected about 5 years ago based on what we thought the ethnic background of the population in Florida consisted of and what the best commercial panel was.

Dr. Farrell recommended that we go back and review the mutations present in CF patients in Florida and see how it compares to our panel; this is doable since the CFF has an IRB approved informed consent data registry. Most CF patients in Florida are enrolled in this patient data registry. This registry does collect mutation data.

Once we have data on the mutations in Florida, Convene a panel (CF center directors, nurse specialists, staff from the lab and CMS employees) to make recommended changes

He also recommended that we obtain information from the CFF to determine our false negative rate.

When NBS was set up, they omitted the requirement to report kids diagnosed after NBS; that were missed through NBS, so we do not know what our false negative rate is. It may be helpful to obtain data from CFF to determine false negative rates

Rapid communicating of results to families; most centers do this very well, although there are a few that are lagging behind a bit.

Recommended reporting to NBS should be required of all children diagnosed with CF through other means(missed by NBS) to get a handle on false negative rates

Recommended on-going meetings or phone conferences twice a year with lab staff, representatives from CMS and CF centers

Recommended that we formally renew contracts with the CF centers, which goes over the original requirements of the contract, in terms of how rapidly we contact families, how rapidly we get sweat tests done, and how rapidly we close cases.

The next steps after the review included a phone conference to discuss recommendations has already taken place; additional meeting planned for October 2012.

Dr. Hudak has put in a data request with the CFF for the data registry to look at the mutations and pick up kids missed by NBS; awaiting approval from Nemours IRB (hopefully soon)

CF centers have been sent letters of agreement with Quality Assurance measures

Work with DR. Farrell once the data from the CFF has been obtained;

Convene panel to review data

Maybe involve geneticists in the panel as well, if anyone is interested

Look at mutations panel to see if any adjustments need to be made

See how many kids were missed; find false negative rate and monitor this going forward

Monitor CF centers to make sure goals are being met

SCID Implementation – Panel discussion with Jojo Dy and Elena Perez(via conference call)

Jojo Dy discussed the implementation of SCID this year

SCID is a genetic disorder of the immune system that occurs when there is a severe defect of T cell production and function.

Why are we testing for SCID? There is an available method to test for SCID and is treatable by stem cell transplant. Early diagnosis and treatment (95% success rate if done by 3.5 months) the success rate goes down drastically to about 66% success rate if done later than 3.5 months. The key to survival is early diagnosis and treatment; it is imperative.

May 2010 – United States Health and Human Services Secretary's Advisory Committee on Heritable Disorders in Newborns and Children added SCID to the Recommended Uniform Screening Panel

January 2011 – Florida Genetics and Newborn Screening Advisory Council unanimously voted to add SCID to the NBS panel

The request for spending authority to implement SCID was vetoed by Governor Scott in 2011.
April 2012 – approved with budget authority to test for SCID

Implementation plan –Contract PerkinElmer services to provide personnel, instruments, reagents and specimen testing. All personnel will have Florida license and tests performed under Bureau of Public Health Laboratory (BPHL) license. NBS staff will QC assays and final approval of results.

Provide PE with work space and other requirements

The SCID algorithm has already been developed with different determinations and recommendations. Perkin Elmer and the CDC will be providing the training. The lab will be performing validation and proficiency testing. They will be submitting the IRB review application for exemption (non- FDA approved)

First test performed will be the TREC – marker for SCID testing - All values are tentative

Anything above 40 is considered normal and will be reported as normal

= or less than 40, will repeat test with beta actin

A lot of testing decisions are based on whether baby is pre-term or full-term (full term being at least 37 weeks)

If baby has previous borderline, it will be treated as a positive

Mailer texts will include the following:

Normal – within normal limits

Abnormal (presumptive positive) - The result suggests that the infant may be at risk for Severe Combined Immune Deficiency (SCID) or other immune deficiency disorders

Recommended action: This infant has been referred by CMS to a specialist for further diagnostic testing. Repeat specimen is not required. For further assistance, contact CMS at 866-804-9166.

Borderline - A borderline elevation of TREC suggests that this infant may be at risk for an Immune deficiency disorder

Recommended action: A repeat dried blood specimen must be submitted IMMEDIATELY to the Newborn Screening Laboratory in Jacksonville. For further assistance, contact CMS at 866-804-9166.

Low - A Low TREC result may be seen in samples from babies less than 37 weeks of gestational age.

Recommended Action: When the baby's age is equivalent to or greater than 37 weeks of gestation, IMMEDIATELY collect and submit another specimen to the Newborn Screening Laboratory in Jacksonville. For further assistance, contact CMS at 866-804-9166

Inconclusive - The INCONCLUSIVE result is due to amplification failure during testing.

Recommended action: When the baby's age is equivalent to or greater than 37 weeks of gestation, IMMEDIATELY collect and submit another specimen to the Newborn Screening Laboratory in Jacksonville. For further assistance, contact CMS at 866-804-9166.

Based on California's experience

- Babies screened 370,000 (FL – 214,000)
- Non-Normal Results: 285 (165)
- Flow Cytometry: 43 (21)
- Referred
 - NICU: 241 (139)
 - Flow cytometry: 28 (17)
 - Repeat screening: 213 (123)
 - Non-NICU: 44 (26)
 - Flow cytometry: 15 (9)
 - Repeat screening: 29 (16)

TREC is low in premature babies

Expecting 165 non normal results

Repeat screening for NICU babies will be high

confirmed cases

- SCID: 5 (TREC: 0)
- SCID variants: 6 (TREC: 0, 4, 6, 10, 20, 25)
- Non-SCID T-cell lymphopenias: 3 (0, 16, 19)

Prevalence rates

- SCID and SCID variants: 1:34,000 births (6)
- T-cell lymphopenias: 1:26,000 (8)
- Hispanic SCID and variants: 1:26,000 (8)
- Hispanic T-cell lymphopenias: 1:20,000 (11)

In California, 5 babies confirmed with SCID, all 5 with a TREC level of 0

Heather Smith (SCID Angels for Life) added – There are many challenges ahead, babies are still being born and need testing. Move forward full force with the tools that we have. Funding came through July 1, so please move as quickly as possible. We have two centers that can do transplants, we should be lucky to have that ability

Dr. Perez will reach out to UM about transplants and will report back to the group.

Dr. Pitel stated that we have precedent with the CF centers with a mechanism in place. He spoke about how great it is that we are so far; leaps and bounds from January

Newborn Screening Update – Lois Taylor

Lois Taylor shared updates, statistics and numbers for the Newborn Screening Program:

1. MSMS borderlines continue to rise each year. We will be addressing these issues with lab; there is a meeting scheduled in August to discuss with the lab.
2. Lois is participating in a neonatal substance abuse work group to find out if there is any correlation to MSMS; so far, no correlation found.
3. Total babies diagnosed – 10% increase from 2010 to 2011 – we did not add a disorder. T4 and TSH screening may have an effect. The numbers are expected to increase even more (30-40 babies) due to open case reports for Sickle cell and CF.
4. New information in packet - Graphs and charts. The first graph shows FNSR tracking vs. Physician requests and number of phone calls made by Newborn Screening Follow-up staff.

Online access implementation lowered phone calls and increased online access.

Phone calls staff has processed Shows that we get a lot of response from letters.

Hearing follow up makes a lot of phone calls to contact families and Audiologists

5. Ms. Taylor discussed the other handout in the packets. This handout showed Hearing data. The hearing data is getting better.

Ms. Tempson added – These are the initial screenings shown on this graph. The areas that we struggle with are repeat testing and follow-up. Lost to follow-up is our issue.

Dr. Fifer added that NICU babies have so many other appointments and hearing is so low on that list. Dr. Fifer reiterated the need for a Spanish speaking individual to speak with families that have Spanish as their first language. 1-3-6 rule is a great guideline but should not be a hard fast rule. We should strategize on how to close the gap on not screened NICU babies.

Ms. Tempson added that the babies that are not screened due to being in the NICU are not included in this number. 2,951 babies from 2011 are still showing up as not screened – NICU.

6. CCHD – CMS did submit a BIP to implement CCHD; advisory council recommended to add it to the panel. We do not need legislation for CCHD because it has been recommended by the advisory council. However, DOH does need funding to implement the screening. Funding has been requested for modification of the data system and follow-up staff (3 additional staff for phone calls). BIP is for \$204,922. CCHD would not be added to the blood spot card, because it is full.

Hearing Screening Update - Pam Tempson

Ms. Tempson showed the “Loss and Found Video” customized for Florida. The purpose of the video is to show the importance of follow-up for families thinking of getting pregnant, are pregnant, or whose baby has not passed the newborn hearing screening. We have the rights to the video. We will post on website, email to all distribution lists, ask that they show in their waiting rooms, show to families, however they can utilize it.

Of the 213,000 live births in 2011, 206,000 screened for hearing. 6500 babies did not pass their initial hearing screen. We are still following up on almost 1000 babies from 2011. Before closing a case as “lost to follow-up”, the baby must be at least 6 months old, we send at least three letters, attempt to contact the family at least three times, and we call and fax the infant’s pediatrician on file. We also use other sources to obtain contact information for the family. Of the babies born in 2011 that did not pass their initial hearing screening, approximately 200 babies were diagnosed with a hearing loss, 65 were closed as lost to follow-up, 32 declined follow-up testing and 14 babies passed away.

Ms. Tempson showed data on babies diagnosed with a hearing loss and how many are enrolled in early intervention services. She noted that the numbers are low for 2010 through 2012, as these babies are still being diagnosed and reported to the state.

Overall, the hearing loss prevalence rate from birth to three fluctuates between .11 and .14%.

Ms. Tempson discussed the Hearing Program’s accomplishments. We are moving along with eReports, which Drew Richardson will discuss. We have started using Direct Secure Messaging (DSM), which is a secure way to electronically send information. All of the hearing contacts have been enrolled. Monthly reports were emailed via DSM in June. We are making progress with 1-3-6 goals. We are making steady progress from year to year and this has been the result of all EHDI staff working hard, from the screeners at the hospitals, the outpatient audiologists, to the service coordinators with Early Steps. We continue to play catch up with follow-up of babies who did not pass their newborn hearing screening and subsequent results have not been received. We still have about 900 2011 babies in addition to a growing number of 2012 babies that are still pending. We are further along than we were at this time last year, but we have a lot of work yet to do.

Monthly rewards continue to be provided to top performing providers among birthing facilities. We will soon be starting to award audiologists and Early Steps programs for their excellent performance as well.

We continue to encourage audiologists to become CMS-approved. These are highly qualified audiologists to whom birth facilities and physicians can appropriately refer children who fail newborn hearing screening or those young children with hearing concerns.

Ms. Tempson provided our contact information and introduced Laura Olson, our hospital educator.

The issue of billing and parents refusing the hearing screen due to the fear of high cost was discussed. Ms. Vickers stated that she and Ms. Taylor will elevate this to legislative planning and general counsel.

Newborn Screening technology updates—Drew Richardson

Mr. Richardson discussed 3 projects he is currently working on. eReports is a web interface funded from a federal grant. eReports is accessible by internet. The user has the ability to validate data before submitted. The submitted data will feed directly into our system. The server was installed and we are looking for implementation mid fall 2012. Live data testing will occur in the next few weeks. These screens are meant for hearing. There will be two screens, one for hearing screenings and one for diagnostic testing.

The other project Mr. Richardson discussed is ELO\ELR(Electronic Lab Ordering/Electronic Lab Reporting). Electronic format to send and receive data.

Some modifications will need to be made by hospitals in a data packet. This would eliminate data entry on all sides. This will increase lab's ability to bill due to reliability of information. This data transfer would occur prior to the arrival of the blood spot card to the lab. This is a much more efficient way to send and receive information. Reliability is a huge benefit. This will also assist the lab in its ability to bill.

This is a massive project that will take years, and it is possible that some of the smaller hospitals may never use this system. There may be little to no return on investment for small birthing hospitals. We will start with the largest birthing hospitals and the hospitals that respond that they would like to participate.

The third project Mr. Richardson discussed is Direct Secure Messaging (DSM). DSM is an internet based secure email. It is self-contained; you cannot email into it and you cannot email outside of it. Medical professionals can communicate about patient information. Currently, all referrals are being faxed. Faxes are being lost, aren't going through, etc. With DSM, you get confirmation that the email went through. It is more efficient and effective.

All genetics centers have been set up to receive referrals via DSM.

New Business – Paul Pitel

Dr. Pitel discussed the need to go to legislature to amend the Newborn Screening Statute regarding GNSP members. We will need a representative for SCID, someone with expertise in the area. It is critical to make this happen.

Ms. Taylor noted – We can put a SCID expert in an open spot next time there is a vacancy. Looking forward, we will want someone on the council representing SCID and someone representing CCHD. It may be beneficial to expand the membership to two practicing pediatricians. Can it be added to the CCHD BIP? If you open up the statute for this, then you open it up to all to change. We will need to request expert advice on this subject.

Dr. Pitel moved on to the next item for discussion, Confirmatory testing laboratories. Dr. Bodamer is merging his lab with Miami Children's lab to create a larger lab. This will improve time lag for confirmatory testing. There is currently a database for two counties to show results; they are hoping to expand to other counties. Labs could be shipped via Fedex. Anyone can use the Miami lab.

All Children's has a lab, but they are limited to testing Amino Acids.

Dr. Bodamer has emailed hospitals to let them know they are available

There are huge benefits from using this lab. Create a working group within the council to identify problems and solutions with lab testing. Dr. Bodamer and Dr. Pitel will join the group; also will need a USF representative and Nemours representative.

Create a working group to report to the council on the following areas:

Confirmatory testing labs

Database piece – capture all babies and track them long term

Screening and confirmatory testing algorithms

Obtain uniform definitions on confirmed disorders
Fifer motioned
Unanimous

CCHD – Gul Dadlani and Jeff Jacobs

Andrea Pilna shared her story about her son who had passed away from CCHD.

Dr. Dadlani shared an overview of CCHD. Cardiovascular disease is present in all stages of life. Pulse oximetry is different than the screenings for most Newborn Screening disorders. Approximately 40,000 children are born with CCHD each year in US. Today, about 90% of these babies will reach adulthood, so adult congenital heart disease is becoming the highest incidence of heart disease.

Children will leave the hospital with CCHD undetected, the PDA closes at one week, this is when symptoms will show. We know that children are leaving the hospital with congenital heart disease and will start showing symptoms within 1-3 weeks of age.

In 2011, the American Academy of Pediatrics recommended a strategy to implement pulse oximetry screening in hospitals. In order to eliminate false positives, they recommended that the test be performed after 24 hours of age. They also recommended the pulse ox be placed on the right hand and either foot. Vital signs are already being taken every 4 hours in newborn nurseries; it just needs to be recorded. It would just need to be determined how it is going to be reported to the state.

Total of three screens before an abnormal is determined, unless <90%, then it will be automatically be abnormal. Pulse ox can detect many defects that could be missed.

Jeff Jacobs called in to speak about the CMS cardiac subcommittee. The subcommittee met Monday July 9th via conference call and unanimously endorsed the implementation of CCHD, decided to make effort for neonatal pulse oximetry to be implemented as a standard.

By October 1st, Pulse ox will be implemented in the 8 cardiac programs/hospitals in Florida. The subcommittee plans to collaborate with the Advisory Council to implement screening at all hospitals. Relationships with all hospitals would need to be made to assure small hospitals have a referral center for echocardiograms.

In order to make this happen, the committees will need to go to legislature. A joint subcommittee with members from both committees should be created to advocate through legislature. They will also need family advocates to tell their stories and make this personal.

Mr. Jacobs stated that he will contact Bill Neely in Atlanta who is working in Washington DC at the National level. He will be a great resource.

James Mosteller with the American Heart Association added that the AHA has their summer Heart heroes program. Some parents that are members have had losses as well. Mr. Mosteller will ask to see who is interested in telling their story. It would also be beneficial to find out the financial difference between scheduled infant heart surgery versus emergency infant heart surgery.

Dr. Dadlani added - Some of these numbers are already in AAP articles

James Mosteller stated that he will use similar language to what he did last year.

Associations need to put this on their list of priorities and get everyone on board.

We should submit recommendations from our advisory council and one from cardiac advisory council.

We will use eReports to report CCHD/pulse ox results. The Budget Issue Proposal to add CCHD testing to the NBS panel is being prepared for consideration by State Surgeon General. The state will want all results reported. We are now currently waiting on DOH to look at the proposal and either submit or reject

Dr. Pitel called for a vote on the following – take the language on CCHD from January, update/modify slightly and send out.

Dorothy Shulman seconded, adding that we should make this as efficient as possible for follow up staff. NBS would just be gathering the information for reporting and data/statistics. Some type of reporting database should be created.

All in favor

Jeff Jacobs added that he will help in any way possible with the cardiac sub committee

Public Comments

There were no public comments.

Minutes approval

The minutes from January 2012 were approved

Adjournment

The meeting adjourned at 2:40pm EST.