

GENETICS AND NEWBORN SCREENING ADVISORY COUNCIL MEETING

The Genetics and Newborn Screening Advisory Council meeting was held on Friday, February 6th, 2015 at the Florida Department of Health (DOH) Bureau of Public Health Laboratories, 1217 Pearl St, Jacksonville, FL.

Call to Order:

The meeting was called to order at 10:04am EST by Paul Pitel, MD, Chairperson of the Council. Roll was taken and introductions were made.

Members Present:

Paul Pitel, MD, Chairman, Jacksonville
Roberto Zori, MD, Gainesville (UF)
Robert Fifer, PhD, Miami (UM)
David Auerbach, MD, Orlando
Dorothy Shulman, MD, Tampa (USF)
Bonnie Hudak, MD, Jacksonville
Keith Nash, March of Dimes, Orlando
Cyril Blavo, DO, MPH & TM, Ft. Lauderdale (Nova)
John Waidner, MD, Jacksonville
Carina Blackmore, MS Vet Med, PhD, Tallahassee
Celeste Philip, MD, MPH, Tallahassee
Melissa Perez, Tallahassee (via teleconference)
Heather Smith, Lakeland (via teleconference)
Lori Gephart, RN, APD, Tallahassee (via teleconference)

Visitors:

Susanne Crowe, MHA, Jacksonville
Jeanne Brunger, PerkinElmer, Inc.
Shana Bauer, Audiology Extern, University of Miami
Chelsey DeFour, Audiology Extern, University of Miami
Larry Vroegindewey, DOH SCID
Sharon Bowden, Pediatrix Medical Group
Helen Burgess, University of Florida
Cheryl Garganta, University of Florida
Susan Weinger, Pediatrix
Bonita Taffe, PhD, MPH, Bureau of Public Health Laboratories, Jacksonville
Lindsay Raub, University of Florida
Kathryn Kinym Munck, Nemours
George Fox

DOH Personnel Present:

Lois Taylor, RN, CMS, Tallahassee
Whitney G. Jones, CMS, Tallahassee
Drew Richardson, CMS, Tallahassee
Pam Tempson, CMS, Tallahassee

Marna Zok, RN, CMS, Tallahassee
Cassandra Pasley, CMS, Tallahassee
Ming Chan, PhD, Bureau of Laboratories, Jacksonville
Patricia Parrish, Bureau of Laboratories, Jacksonville
Sherry Ray, Bureau of Laboratories, Jacksonville

Conference Call:

Donna Barber, RN, CMS, Tallahassee
Bobbie-Jean Armstrong, RN, CMS, Tallahassee
Sue Meter, RN, CMS, Tallahassee
Emily Reeves, RN, CMS, Tallahassee
Elaine Grace, RN, CMS, Tallahassee
Linda Deterding, RN, CMS, Tallahassee
Donald Fillipps, MD, University of Florida
Representative Michael La Rosa

Housekeeping/Reminders

Dr. Pitel reminded council members and guests to hand in their money for lunch and to review the minutes of the July 2014 council meeting for approval after the break. Dr. Pitel suggested to the council that a new process for approving minutes two weeks after each advisory council meeting be implemented for the council. All members of the council agreed and the motion passed unanimously.

New advisory council member Dr. Carina Blackmore introduced herself to the council as the Bureau Chief of the Bureau of Public Health Laboratory. She has a master's degree in Veterinary Medicine and a PhD.

Dr. Bonita Taffe and Sherry Ray both introduced themselves as new members of Newborn Screening staff at the laboratory. Cassandra Pasley introduced herself as the new Division Director of Children's Medical Services.

2015 Legislative Update

Dr. Celeste Philip gave a legislative update to the advisory council. SB 146 (Companion HB 23) is related to "Screening for autism spectrum disorder". This bill is proposing that physicians will use American Academy of Pediatrics' guidelines to determine if a referral to a specialist is necessary or a parent can self-refer to Early Steps or to another specialist in autism. Specialists will be required to have training in validated diagnostic tools. This bill will require health insurance plans and health maintenance contracts issued or renewed on or after January 1, 2016 to provide direct patient access for a minimum of three visits per policy year. This proposed bill could significantly increase the number of referrals to the Children's Medical Services Network and Early Steps programs for children requesting screening who have a diagnosis of autism spectrum disorder and are in need of early intervention services.

HB 403 was presented by Representative La Rosa to add Newborn Adrenoleukodystrophy (ALD) Screening to the newborn screening panel. This bill will require ALD screening within 24 hours of age and physicians to be responsible for ALD screening for birthing centers and home births. It does provide for parents to have a right to decline the screening. The group discussed the costs associated with this bill and methods of diagnosis.

Cystic Fibrosis (CF) Data

Bonnie Hudak gave a presentation to the council regarding Cystic Fibrosis data. The Florida Department of Health contracts with accredited Cystic Fibrosis centers in the state of Florida to provide a diagnostic evaluation for babies with an abnormal cystic fibrosis newborn screening result. The Cystic Fibrosis Foundation has a quality improvement consortium with representatives from all over the states and collects data from different states to compare timeliness. The Foundation looked at infants diagnosed between 2010 to 2012 (not including prenatal diagnosis). Florida is fifth in the US in terms of infants diagnosed with CF. The goal is to have infants diagnosed before 15 days of age and Florida is diagnosing between 13 to 14 days of age and is meeting the CF Foundation guideline. The median age for first visit is around 40 days and it is unknown why there is a lag between diagnosis and first visit. Some of the CF referral centers in Florida expressed concerns with Managed Medicaid Programs that are requiring prior authorization before obtaining a sweat test for diagnosis. Dr. Hudak asked the council if it would be appropriate to draft a letter to send to Agency for Healthcare Administration (AHCA) urging them to work with the managed care programs to try and bypass the prior authorization for newborn screening diagnosis. Dr. Hudak showed an educational video for CF parents to the council that was paid for by the Cystic Fibrosis Foundation. The hope is that this video will be distributed nationwide through the CF Foundation. Dr. Hudak and her team are in the process of making a video to distribute to providers in Florida.

Dr. Fifer made a motion for the council to draft a letter to send to ACHA regarding the prior authorization issue. Dr. Auerbach seconded the motion and the council vote was unanimous. Dr. Pitel suggested that Dr. Hudak draft the letter and Dr. Pitel would sign it on behalf of the council. Dr. Philip stated she would reach out to AHCA to get some information regarding the managed care programs and newborn screening.

NewSTEPs Report Recommendations

Lois Taylor presented the recommendations made by the NewSTEPs report to the council for consideration. A survey of all fifty states showed that Florida is one of only five states that still required the 24 hours of protein feeding. Dr. Cheryl Garganta stated that with the new technology of Tandem Mass Spectrometry testing the sensitivity of the test is better and therefore the 24 hours of protein feeding is less critical. The downside to dropping this requirement is to amend the format of the specimen cards but it is necessary as it would reduce unnecessary repeat testing. The council was unanimously in favor of accepting this recommendation.

The NewSTEPS report also made a recommendation for an updated storage and retention policy. The report suggests that blood spots should be kept longer and be used for other purposes. Dr. Pitel stated that Florida does not request consent from parents to use blood spots for other purposes besides newborn screening and this presents a challenge. To use blood spots for research purposes is not possible under the current Florida statute. Patricia Parrish stated that if the blood spots were to be kept longer that it would require frozen storage and there is not enough freezer space for longer retention. Dr. Bonnie Taffe stated that if the blood spots are to be used for research purposes then all specimens would have to be put in freezer storage. The council agreed that more information should be gathered on the retention policy for quality assurance and future research possibilities before making a decision on this recommendation.

Genetic Centers

Dr. Roberto Zori gave the council an update on the future of Florida's Genetic Centers. The role of Genetic Centers is to treat all metabolic conditions of which most are picked up by Newborn Screening. As the Newborn Screening Program has grown the funding for Genetic Centers has stayed the same. There is a large waiting list for patients at the genetic centers. Many of the patients are adults now and there is no reimbursement because 90% of the work with these patients is done by phone. The money given to the genetic centers this year is providing help to survive as well as look at other ways to provide services efficiently. Telemedicine is a way to provide services across the state efficiently as well as give patients the choice in who is serving them and eliminates their need for travel. The council discussed the benefits of creating a telemedicine network for the genetic centers. Dr. Philip asked Dr. Zori to provide written information on how the genetic centers operate, statistics on savings in terms of travel, and costs associated with the families served. Dr. Zori agreed to work with DOH to compile this information.

The advisory council approved the July 2014 minutes with minor suggested edits.

Representative La Rosa spoke to the council about Adrenoleukodystrophy (ALD) and HB 403 that he is sponsoring. Two years ago a family approached Rep. La Rosa regarding ALD and the possibilities for a screening for ALD. Last year a bill was filed to add ALD to the newborn screening panel but was not passed. This year HB 403 is attempting a similar result to add ALD. ALD is coming up for review at the federal level to add ALD screening soon. Rep. La Rosa would like Florida to lead the way. La Rosa suggested that he meet with the council later in the year to discuss ALD and newborn screening. Dr. Pitel expressed his gratitude to Rep. La Rosa for his interest in newborn screening. Dr. Pitel stated that Florida has adopted and implemented all federal recommended screenings. Dr. Garganta stated that it may be possible to add screening for ALD to the panel but follow-up afterwards would be difficult. Screening for males is easier than screening for females since females would require more in depth analysis testing. ALD cannot be distinguished between early-onset and adult-onset and therefore treatment is difficult. Dr. Zori suggested that before adding a new disorder to the screening panel it needs to be determined how the program is doing right now. The program needs evaluated for strength before adding anything new. The advisory council spoke about having a

presentation done to outline the requirements for testing and treatment of ALD at the next advisory council meeting.

Newborn Screening Laboratory Update

Dr. Bonnie Taffe gave the advisory council an update on the Newborn Screening Laboratory activities. Current vacant positions are the NBS Lab Director, a Chemist Administrator, and three Chemist II positions. New staff is Dr. Bonita Taffe who is a Chemist Administrator and Acting NBS Lab Director. Jasmine Brown was hired as a Chemist II. Over the years the lab has decreased in size from 28 staff members to 19 staff members. There are a few staff who are close to retirement.

Quality Assurance proficiency testing achieved 100% in the third and fourth quarters in 2014 for all areas of hemoglobinopathies, Cystic Fibrosis, Severe Combined Immunodeficiency (SCID), GSP (endocrine and enzymatic disorder), and MSMS (amino acid and fatty acid disorder). Dr. Cheryl Garganta has contracted with the state lab to provide input on improving lab procedures and has visited the lab for consultation 8 times since August 2014.

Screening Center went live on October 13, 2014. Screening center is a software upgrade for LifeCycle. There were many errors, corrections, and delays with the implementation but most issues are now fixed and the list of items has been significantly reduced. Currently the lab is current with data entry of demographics.

Newborn Screening Follow-Up Program Update

Lois Taylor gave the advisory council an update on the Newborn Screening Follow-Up Program. The NewSTEPs evaluation was the third evaluation of the Florida Newborn Screening Program by a national review team. NewSTEPs asked us to provide feedback to them about what suggestions were or are to be adopted by the NBS program.

NewSTEPs recommended that the list of screening disorders be incorporated into the Newborn Screening rule so that the rule need not be updated every time a new disorder is added to the panel and that recommendation has already been implemented. Unfortunately the document that is incorporated by reference would need to be revised each time the list is updated.

The unsatisfactory specimen rate decreased 67% from 2.36% to 1.60% because the Follow-up Nurses were able to visit hospitals in 2014 (none went out in 2013 due to staff shortage). Hospitals submit 96% of all specimens and 80% of the unsatisfactory specimens but the other submitters who submit only 4% of specimens account for 20% of the unsatisfactory specimens. The birth rate is slightly up but the number of specimens is decreasing which shows that educational and Follow-up efforts decrease un-mandated repeats.

In 2013 there was a false elevation of the number of SCID referrals due to one hospital using heparinized lines when collecting bloodspots. The NBS Follow-up nurse assigned to the

hospitals in the North East part of Florida worked with that particular hospital to educate them on correct collection methods. Recent analysis in August showed the difference after education efforts were provided in January.

2013

SCID Inconclusives - **35** out of 281 statewide were from Hospital X

SCID Early - **7** out of 48 statewide were from Hospital X

SCID Borderline - **4** out of 28 statewide were from Hospital X

SCID PrePos - **13** babies were screened as PrePos from Hospital X and 0 were actual SCID

2014

SCID Inconclusives - **18** out of 165 statewide were from Hospital X

SCID Early - **0** out of 5 statewide were from Hospital X

SCID Borderline - **0** out of 5 statewide were from Hospital X

SCID PrePos - **0** babies from Hospital X were screened as PrePos

GALT and Biotinidase referrals have increased. Dr. Garganta who is contracted with the NBS Lab as a consultant will be looking at the increase of MS/MS borderlines. Printing of Sickle Cell Trait letters, Unsatisfactory Specimen letters, and Hearing letters have decreased from last year – probably due to the lower specimen unsatisfactory rate and the Hearing Follow-up Program catching up on the outstanding cases..

Pompe Disease was recommended by the Secretary's Advisory Committee for Inheritable Disorders for Newborn Children (SACHDNC) about two years ago but was never added to the Recommended Uniform Screening Panel by the Secretary of the US Health and Human Services. Instead she ordered an independent report but it is not yet available.

Adrenoleukodystrophy (ALD) is to be reviewed for addition to the Recommended Uniform Screening Panel. The Advisory Committee will be meeting in February to address ALD and other issues.

The D/SACHDNC Laboratory Standards and Procedures Subcommittee is compiled a report with suggested recommendations for timeliness in newborn screening which will be presented to D/SACHDNC in February 2015 for final approval.

The suggested recommendations are:

1. Presumptive positive results for time-critical conditions should immediately be reported to the child's healthcare provider and no later than 5 days of life.
2. All presumptive positive results for time sensitive conditions should be reported to the healthcare provider as soon as possible but no later than 7 days of life.
3. All NBS results should be reported within 7 days of life (the "normal" screening results).
4. In order to achieve these goals (and reduce delays in newborn screening):
 - Initial NBS specimens should be collected in the appropriate time frame for the baby's condition but no later than 48 hours after birth.

- NBS specimens should be received at the Laboratory as soon as possible; ideally within 24 hours of collection.

Ms. Taylor asked the council for a recommendation to change Florida's reports for timeliness from 5 days to 3 days. Right now the statewide average for specimens arriving at the lab is 2.78 days. The national standard is within 24 hours from specimen collection. The council discussed the issues of penalizing offenders who do not meet the standard. The council recommended that the program proceed with changing Florida's standard from 5 days to 3.

Critical Congenital Heart Disease (CCHD) data began being collected July of 2014. Of the 43 babies that have failed the CCHD screening, 3 were diagnosed with CCHD. Two babies were diagnosed prenatally.

The Newborn Screening Program began in Florida in 1965. This year Florida's Newborn Screening Program will be celebrating its' 50th anniversary. Plans are to create materials in celebration of this anniversary and create posters to send out to physicians in Florida.

Newborn Hearing Screening Update

Pam Tempson gave the council an update on the Newborn Hearing Screening Follow-Up program. Dr. Charles William of the University of Florida recently resigned as Florida's EHDI Pediatric Champion and recommended Dr. Donald J. Fillipps as his successor. Dr. Fillipps is a Clinical Associate Professor with the Divisions of General Pediatrics at the University of Florida. The role of the EHDI Pediatric Champion is to serve as the point person on newborn hearing issues at the state and local levels to ensure success of reaching each of the American Academy of Pediatrics' (AAP) EHDI program goals. Each state has an EHDI Pediatric Champion that was appointed by the AAP.

eReports™ now has 255 registered users who are entering the majority of hearing screening data that was previously faxed and entered by CMS follow-up staff. As of February 6th all hospitals have been trained in the use of eReports™ for reporting hearing screening data.

Of the 207,782 babies screened in Florida in 2013, 199,749 passed the initial screening and 7,991 did not pass the initial screening. Of those 7991 that did not pass the initial screening, 6726 passed a subsequent test, 254 were diagnosed with a permanent hearing loss, 770 were lost to follow-up, 19 cases are still pending, and the rest were closed due to various reasons. Data shows that since 2009 the percentage for lost to follow-up cases has decreased from 22.59% in 2009 to 9.56% in 2013.

eReports™ , ELO/ELR, Screening Center Update

Drew Richardson gave the advisory council an update on Newborn Screening systems. Florida Newborn Screening Results (FNSR) has been live since 2009. Currently there are 4,133 users of this program and it is processing around 95% of Newborn Screening lab report requests. Access to this program was expanded in June of 2014 to include new medical

licensures such as audiologists, etc. A new Kit Number (unique blood card identifier) search field should be coming soon for this program.

eReports™ has been live since June of 2013. eReports™ allows audiologists to electronically report results to the program which significantly decreases faxing results. About 80% of hearing screening data is entered through eReports™. Currently there are 276 trained users of this system. As of February 2015 all hospitals have been trained in the use of this system.

Electronic Laboratory Ordering\Electronic Laboratory Reporting (ELO\ELR) project started back in 2011. There are currently 11 hospitals working on this project and three of those hospitals have assembled project teams. Earliest adopter of this project is likely to come in late spring of 2015, so the hopes are to have more information to the advisory council on this at the next meeting.

Screening Center is the new laboratory information management system. Screening Center went live on October 13, 2014.

Move IT is a system the Newborn Screening Program used for electronic transfer of protected health information to and from referral centers. Using this system was necessary because DOH did not have an encrypted email system to send information securely. As of yesterday, February 5th, Florida Department of Health's email system now has encryption capabilities.

New Discussion Items

Dr. Fifer inquired about the bill passed last session requiring audiologists to check with parents to see if they wanted education materials sent to them regarding hearing loss. It has been suggested that there are some providers on that list that were not appropriate to be on there, such as providers who supply hearing aids. Lois Taylor stated that only Early Steps providers are on that list and therefore all of those providers are approved to send information to parents if requested. Since hearing aids is considered assistive technology it fits within the rules of the bill.

Dr. Shulman stated that previously there were talks regarding doing de-identified research such as the incidence of congenital hypothyroidism or children identified with an abnormal pulse oximetry screening. Is it possible to review de-identified data for research? Dr. Philip suggested looking at proposing an IRB to request that data for research.

Adjournment

The meeting adjourned at 2:19pm EST.