GENETICS AND NEWBORN SCREENING ADVISORY COUNCIL MEETING

The Genetics and Newborn Screening Advisory Council meeting was held on Friday, July 31st, 2015 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 10:13 am EST by Paul Pitel, MD, Council Chairperson. 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, St. Petersburg (USF)
Bonnie Hudak, MD, Jacksonville
Carina Blackmore, MS Vet Med, PhD, Tallahassee
John Waidner, MD, Jacksonville
Elena Perez, MD, Miami (UM)
Keith Nash, March of Dimes, Orlando
Melissa Perez, Consumer, Tallahassee
Heather Smith, Consumer, Lakeland
Lori Gephart, RN, APD, Tallahassee (via teleconference)
Cyril Blavo, DO, MPH & TM, Ft. Lauderdale (Nova) (via teleconference)

Guests:
Jeanne Brunger, PerkinElmer, Inc.
Christian Gentile, Audiology Extern
Sierra Lawrence, Audiology Extern
Brittany James, Audiology Extern
Sharon Bowden, Pediatrix Medical Group
Scott Williams, Baebies
George Fox
Barry Byrne, MD, PhD
Rodney Howell, MD, FAAP, FACMG
Kari Morgenstein
Gerald Shiebler, MD
Ashley Cooley, MD

DOH Personnel Present:
Whitney G. Jones, CMS, Tallahassee
Bonita Taffe, PhD, MPH, Bureau of Public Health Laboratories, Jacksonville
Larry Vroegindewey, DOH, SCID
Ming Chan, PhD, Bureau of Laboratories, Jacksonville
Sherry Ray, Bureau of Laboratories, Jacksonville

Donna Barber, RN, CMS, Tallahassee (via teleconference)
Sue Meter, RN, CMS, Tallahassee, (via teleconference)
Emily Reeves, RN, CMS, Tallahassee (via teleconference)
Bobbie Jean Armstrong, RN, CMS, Tallahassee (via teleconference)

2015 Legislative Update

Lois Taylor gave the advisory council an update on the 2015 legislative session. Two Newborn Screening (NBS) bills were filed, HB 403 and SB 632, and they were similar bills seeking to add Adrenoleukodystrophy (ALD) to the Florida NBS panel if it is added to the federal screening panel. Both bills did not pass in session.

Dr. Paul Pitel spoke about the money allocated to the three genetic centers when the Florida budget was approved. In 2014 the centers received $200,000.00, but in 2015 the centers were only allocated $100,000.00. Dr. Pitel made a motion to craft a budget that reflects the cost of operating a genetic center to send to the legislature after advisory council review and approval. Dr. Fifer supported the motion, Dr. Shulman seconded. The council passed the motion unanimously.

Guest Speaker: Newborn Screening from the National Perspective

Dr. Rodney Howell presented to the advisory council regarding Newborn Screening for Pompe Disease. Newborn Screening for Pompe Disease was added to the Recommended Uniform Screening Panel (RUSP) in spring of 2015. The assay for the enzyme deficiency in Pompe is straight forward but the wide spectrum of the disease makes newborn screening follow-up complicated. Most patients diagnosed with Pompe will have later onset conditions.

When the Discretionary Advisory Committee on Heritable Disorders for Newborns and Children recommended adding Pompe Disease to the RUSP, it was recommended that large pilot studies be performed before the condition was added nationally. Three pilot states, New York, Wisconsin, and Georgia, are in various stages of implementing the screening. New York State is using MS/MS technology to identify babies with Pompe. Currently they have screened 180,000 babies, had 28 total referrals with low enzyme activities, and have identified three patients who they think are possible late onset or carriers of Pompe. No infantile cases have been identified through New York State's study. Wisconsin and Georgia are planning to use digital microfluid platform of Baebies which is simpler and cheaper to use than MS/MS. Baebies platform has been judged as research by the FDA, so both Wisconsin and Georgia are on hold until it is resolved.

Prior to launching state wide programs it will be essential to have common data elements in place for long-term follow-up. Newborn Screening Translational Research Network (NBSTRN) has an LSD Workgroup and Pompe Subgroup that has generated common data elements for this purpose. New York and Wisconsin have also generated protocols for short-term follow-up and diagnosis. The Mayo group has also developed modules and will all be available in the near future for sharing.

The Muscular Dystrophy Association and some other smaller groups are in discussions with the NBSTRN about planning some very large pilot studies for Duchene Muscular Dystrophy (DMD). Although there are no specific treatments approved today, there are several drugs currently before the FDA that have the possibility of proving highly effective for the treatment of DMD.

New federal regulations regarding the use of NBS dried blood spots was passed. It states that research on newborn screening dried blood spots shall be considered research carried out on human subjects. NIH funded research using newborn dried blood spots collected on or after March 18, 2015 will be considered non-exempt human subjects research, and therefore, must follow the HHS protection of human subjects regulations. Parental permission must have been obtained in order to use newborn dried blood spots collected on or after March 18, 2015, in NIH funded research.

The council discussed the need to expand the storage of NBS specimens from six months to twelve months. Dr. Pitel suggested a motion to create a working group to discuss the issues and questions regarding expanding blood spot storage to twelve months. The group unanimously passed the motion to create the workgroup. Bonnie Taffe agreed to collect data on the cost of setting up a storage area that has climate control.

Pompe Research Project

Dr. Ashley Cooley presented to the council regarding Pompe Research Project. Classification of Pompe Disease includes infantile and late onset. Infantile Pompe occurs before 12 months of age and death occurs usually within the first year of life or very early childhood. Late-onset Pompe can occur after 12 months of age and most of those with late onset seek medical care for symptoms in adulthood. Many have mild weakness in childhood that can go unrecognized. Diagnosis of Pompe includes establishing low functional GAA enzyme levels and genotyping to rule out pseudodeficiency, identify carriers, predict infantile-onset versus late-onset, and predict Cross-Reacting Immunologic material (CRIM) status. The current treatment is enzyme replacement therapy (ERT) that is initiated as soon as possible and early initiation of ERT improves intermediate term outcomes.

Taiwan currently screens for Pompe disease and both Washington State and Missouri are doing pilot studies on Pompe. Taiwan has screened 470,000 infants for Pompe and 10 infants have been identified by the screening and started early treatment. Washington has identified 4 infants with Pompe, 4 carriers, 3 carrier/pseudo deficiency, and 6 heterozygous for pseudo deficiency. Missouri has screen 43,701 samples with 27 identified to have some type of LSD, 18 screened positive for Pompe, and 8 identified with Pompe (3 infantile, 3 late-onset, and 2 of unknown significance).

The proposed project from Dr. Byrne and Dr. Cooley is to screen de-identified newborn screening samples prospectively for 6 months, confirm with genotyping, and determine incidence and feasibility of the screening process and confirmation process. Dr. Shulman made a motion to explore the effort to pilot a study for Pompe Disease and research the

legality of it. Dr. Pitel supported the motion and Dr. Auerbach seconded. The council passed the motion unanimously. Dr. Byrne agreed to prepare a proposal for the Institutional Review Board (IRB), Pompe Research.

Effects of Antibiotics

Lois Taylor asked the council regarding the bubble on the specimen card for antibiotics. Should babies on antibiotics at the time of specimen collection have a repeat specimen done at 28 days of age? It was indicated that this is a carryover from issues in the past that are no longer valid. Dr. Shulman made a motion to remove the antibiotic bubble on the specimen card. The motion was seconded and was passed unanimously by the council.

The council approved the minutes before moving on to the next topic.

Update on ALD

Lois Taylor updated the council on ALD. ALD was not recommended for newborn screening at the national level. There is not much to update on at this point.

Newborn Screening Laboratory Update

Bonnie Taffe gave the council an update regarding the Newborn Screening Laboratory. There are currently two vacancies at the lab for NBS Laboratory Director and a Chemistry II position. Patricia Ryland is a new Chemistry Administrator who was hired recently, and there are two pending hires for Chemist II positions.

Proficiency of testing is at 100% for the second quarter for hemoglobinopathies, cystic fibrosis, SCID, GSP, and MSMS.

The SCID assay has been FDA approved and transition is being made to the new Automated EnLite TREC kit and instrumentation. This has been budgeted in the 2015-16 fiscal year. A minor upgrade is in progress to improve DNA preparation amplification and analysis of CF mutations and TREC which was a recommendation made by NewSteps. New equipment for SCID is scheduled to arrive and be set up in the fall with training and validation by late 2015 or early 2016. Analysis will transition over to the NBS laboratory staff which will remove the need of PerkinElmer “Lab in Lab” staffing for SCID.

Newborn Screening Follow-Up Program Update

Lois Taylor gave an update to the council regarding the Newborn Screening Follow-Up Program. The 50th anniversary of Florida Newborn screening is this year (2015). There will be a celebration at the capitol on September 24, 2015 and First Lady Ann Scott will be present. The celebration is being held in September because September is National Newborn Screening Month. Governor Scott made an official proclamation of NBS 50th anniversary. Announcements were also made in two magazines, Florida Family Physician and The Florida Nursing Quarterly.

Since January 2015 the Newborn Screening Follow-Up Program has changed. NBS has merged with Early Steps into the new Bureau of Early Steps and Newborn Screening. A nursing supervisor will be hired to supervise the nurses and contract manager of Newborn Screening.

MS/MS Borderlines have seen an increase recently and that is due to a change in the kit used for screening which is more refined. In 2014 the program processed about 12,000 presumptive positive tests and also did about 5,000 follow-up cases. In 2014 CCHD had 43 cases referred for a failed pulse oximetry screening. In 2015 with only 6 months of data there have been 42 cases that have referred screenings.

Dr. Pitel suggested that the newborn screening database be attached to FLSHOTS as this will aid in follow up and also physicians in obtaining newborn screening results.

Update regarding eReports, ELO/ELR, Screening Center

Lois Taylor gave the NBS Technology Update to the council. Florida Newborn Screening Results (FNSR) website has been live since 2009. Currently there are 4,357 users and is processing about 97% of NBS lab report requests. About 20,000 requests are downloaded monthly. A kit number (unique blood card identifier) search was added in March.

eReports has been live since June 2013. 266 end users have been trained to use this system and all birthing hospitals have been trained. About 92% of hearing screening data is entered through eReports.

Electronic Laboratory Ordering/Electronic Laboratory Reporting (ELO/ELR) currently has 13 participating hospitals engaged in various stages of talks for blood card data transmission. The first Go Live is scheduled for Fall of 2015 with Baycare Hospitals and Naples NCH. Tampa General Hospital has expressed interest in the project again.

Screening Center has a couple upcoming enhancements of digital blood card scanning and multi-faceted reporting capabilities (fax, paper, HL-7 data).

New Discussion Items

Melissa Perez stated that some children with Phenylketonuria (PKU) have been denied coverage of medical food through Medicaid/CMS. AHCA has not covered special food for PKU patients. The council discussed this issue and Melissa Perez made a motion to draft a letter to the Agency for Health Care Administration, Dr. Celeste Philip and Dr. John Armstrong of the Department of Health for assistance. Keith Nash seconded the motion and the council passed it unanimously.

Adjournment

The meeting adjourned at 2:20pm EST.