Colorado Newborn Screening Committee

Meeting Minutes

October 18, 2016

Members Present: Anne Behring (by phone) Kay Kelly, Mary Kohn, Michelle Miller (by phone) Suzanne Rogers, Joe Toney, Patrice Whistler (by phone), Lori Wise

Members Absent: Teri Hulett

I. Approval of Minutes

Minutes of the June 28, 2016 meeting were approved by a vote of the Committee.

II. Approval of Bylaws

The Committee bylaws were approved by a vote of the Committee with one addition: add the date enacted as 10/18 2016 and date to be reviewed as 10/18/2017.

III. Changes to the Newborn Screening Program

Tista Ghosh, Deputy Chief Medical Officer and Director of Public Health Programs for the Colorado Department of Public Health and Environment (CDPHE), Ann Hause, the Director of Legal and Regulatory Compliance, laboratory fiscal staff, birth defects epidemiologists, and other representatives of CDPHE (hereafter referred to as the “NBS team”) provided an update regarding trends and changes to the Newborn Screening Program over the past six to nine months. As members are aware, recently the laboratory attempted to add screening for Pompe Disease, an analysis of laboratory finances revealed that funds were not available to do so. The financial analysis then prompted the need to make some changes in newborn screening program processes to align with budgetary constraints. As a result, the department reduced a contract with University Physicians Inc. (UPI) to provide immediate sub-specialist consultation to primary care providers regarding certain newborn screening results. In order to do so, the Department identified “time-critical” results, based on national guidelines, and prioritized maintaining immediate sub-specialty access for those conditions. However, resources in the end did allow for even more conditions to be included in the immediate sub-specialty access category,
including all conditions referred to sub-specialists in the past, with the exception of congenital hypothyroidism. For congenital hypothyroidism, the decision was made to report the abnormal result to the infant’s primary care physician by phone and fax with a copy of the abnormal results and printed information from the American College of Medical Genetics on how to address next steps regarding the result. Contact information for all pediatric endocrinologists (not just UPI endocrinologists) is also provided.

Contracts with subspecialty providers other than UPI (e.g., sickle cell and SCID) continued unchanged.

In addition, CDPHE staff noted that the newborn screening program has had to clarify its role vis-à-vis public health and health care. CDPHE is not a HIPAA-covered entity, and cannot perform functions that are covered under HIPAA. Public health’s role is to prevent disease and to provide screening and linkage to care. Once the linkage has occurred, the health care system’s role is to then diagnose and treat. Previously CDPHE staff was following up to ensure appropriate diagnostic testing and treatment. It was recently determined that this was not within the department’s public health authority, but rather crossed the line into medical care. However, it is within the department’s public health authority to gather information on those who have been diagnosed with conditions through the birth defects registry. Regular reviews of that data may offer information similar to the information gleaned previously through the follow-up phone calls, if infants identified with abnormal screens are diagnosed with disease (true positives).

The NBS team has reviewed national standards and conferred with other states and has determined that Colorado has a higher false positive rate for some conditions. One reason relates to Colorado’s lack of specialized protocols for screening NICU babies, who have high rates of false positives. Using the NICU protocols from other states as a template and comparing to the NBS program’s experience, the NBS team and laboratory scientists will draft a NICU protocol for Colorado. The NICU protocol will hopefully be ready to present to this committee at the next meeting in January.

IV. Staffing Changes

Dan Wright, the Newborn Screening Program Manager, has retired. Erica Wright, the Newborn Screening Follow-up Program Supervisor has resigned. The NBS team is in the process of hiring a Newborn Screening Program Manager. In the meantime, Margaret Ruttenber, the Director of the Birth Defects Registry, is working part-time with the screening lab to temporarily fill some of the gaps left by Dan’s retirement.

V. Fiscal Report

Olga Ivanova, the lab’s Fiscal Services Manager, reported on the NBS program’s financial situation. Three years ago the newborn screening program had a cash fund balance, but an increase to six-day-a-week testing, additional courier services, a yearly increase in supply costs with no corresponding increase in revenue, and other increases in operating costs have significantly reduced those cash reserves, resulting in a negative balance. Funds are very limited for the fiscal year that began July 1, 2016, necessitating a reduction in expenses. Given the steps that have been taken to reduce
expenditures, balance will be restored by the end of the current fiscal year. Revenue can only be increased via a fee increase, the last of which was approved five years ago. Raising the newborn screening fee is not currently an option, given the TABOR amendment and the current state budget.

VI. **Changes in Newborn Screening Procedures**

Ann Hause, the Director of the Office of Legal and Regulatory Compliance at CDPHE, elaborated further upon the difference between public health and health care described above, as defined by HIPAA. Standard operating procedures (SOPs) in the newborn screening program have been devised that eliminate practices that have been in place in the program that are “health care-related.” These SOPs define when the lab contacts the PCP, the sub-specialists, etc. so that all newborns with abnormal screens are connected to a point of care. Advising physicians on diagnostic testing and clinical considerations does not align with the public health role, which is to connect to healthcare, and the medical home. To this end, when abnormal screening results are reported to providers, they are provided with national guidance for next steps and lists of local specialists. The NBS team will also be assessing how well the goals of newborn screening are being met.

Dr. Whistler expressed concern about hypothyroidism. Hypothyroidism is the only condition on the panel that does not involve the notification of a sub-specialty provider. In the past an abnormal hypothyroidism screen would be called out to a pediatric endocrinologist who would locate the appropriate provider and offer consultation on diagnostic testing and clinical care. Dr. Whistler is concerned that it will not be the appropriate PCP on the receiving end of the report of the abnormal screen. If PCPs have infants in their practice they likely know what to do. But locating the actual provider who is caring for the infant can be difficult. Dr. Toney expressed his concern that he has observed instances where information comes to a provider who does not have the patient in the practice and the information is not addressed. Dr. Toney noted that the pediatric endocrinologist would take responsibility for care of that newborn until care could be handed off to the appropriate provider. Dr. Ghosh noted that locating the appropriate provider for an infant has always been a challenge for the program. The NBS team has devised a protocol for assisting in finding and verifying the PCP for this very reason. This assistance is also available to the contracted sub-specialists.

Dr. Barker, a pediatric endocrinologist, said she is aware of more than one instance where the appropriate provider got the information and advised the family to see an endocrinologist, which the family did not do, with no follow-up by the provider.

Currently the PCP is notified of the abnormal result for hypothyroidism and receives the report and national follow-up guidelines. Contact information for the pediatric endocrinologists is provided and PCPs are advised if they have difficulty they can call the NBS program for assistance.

Dr. Toney asked if not contracting with the pediatric endocrinologists is a financial issue. Dr. Ghosh said that is the main reason, which hopefully can be addressed in the future.
Dr. Kohn asked if primary care providers have been educated about this new responsibility. It is important that PCPs know they have the sole responsibility for follow-up of abnormal results for hypothyroidism, that they won’t be contacted by a pediatric endocrinologist for assistance. Dr. Ghosh said the Department can send out information to pediatricians and family practice physicians through the Department’s Health Facilities Portal and other communication vehicles. Dr. Whistler said she thought it was a good idea to notify physicians using various avenues, to reinforce the message. It was noted that there are many categories of providers other than physicians who need to be educated as well.

Dr. Toney asked if parents are called in the case of hypothyroidism. Dr. Ghosh responded that the NBS program has never contacted parents as part of its protocol unless the program has been unable to locate a provider, in which case contacting the parents is a last resort to determine the name of their PCP.

Dr. Sontag suggested that the Attorney General might be presented with the argument that newborn screening is a system, as defined by ASTHO, MCH, and other national bodies. Perhaps that would convince the Attorney General to amend the decision that follow-up is not allowable.

The Committee requested on-going communication and suggested that the NBS team reach out to the sub-specialists for further discussion.

The NBS team at CDPHE raised the possibility of adding both positive and negative diagnostic test results for newborns who have an initial positive newborn screen to the department’s reportable conditions list, through Board of Health rule. That would potentially allow an accommodation for the role formerly played by follow-up phone calls to assist in identifying those who did not receive appropriate diagnostic testing, or who had delays in diagnosis or treatment. It would also continue to allow identification of false positives for quality improvement purposes. Specific language will be drafted for discussion at the next meeting.

VII. Newborn Screening for Critical Congenital Heart Disease (CCHD) - Update
Margaret Ruttenber updated the Committee on the status of newborn screening for CCHD. Between January 1, 2016 and September 30, 2016, 50,930 babies were screened for CCHD using pulse oximetry. There were 241 failed screens. There are many reasons why infants fail a CCHD newborn screen other than CCHD, e.g., trisomy 21, failure to thrive, preterm gestational age, maternal drug use and, most commonly, persistent fetal circulation. The CCHD newborn screening program is designing pilot projects with University Hospital and Children’s Hospital to match failed screens with known outcomes from their facilities. Screening has not been done above 7,000 feet and midwives have not reported many CCHD screenings. Twenty patient charts relative to CCHD have been requested by the registry for January – March of 2016. These records will be reviewed for more detail.

VIII. Public Comment
There were no constituents signed up for public comment.

IX. The meeting was adjourned.