

## **NBS Advisory Council**

*January 16, 2020*

### **Members Present**

Emily Barr  
Carolina Beltran  
Kourtney Bettinger  
Grace Brouillette  
Merlin Butler  
Jennifer Gannon  
Selina Gierer  
Shobana Kubendran  
Michelle Leeker  
Michael Lewis  
Karey Padding  
Deborah Stern  
Meghan Strenk  
Julie Wellner  
Britton Zuccarelli

### **Members Absent**

Laurie Gwyn  
Thomas Loew  
Susan Pence

### **KDHE Staff Present**

Kinsey Anderson  
Michelle Black  
Kayzy Bigler  
Dustin Caldwell  
Jan Conklin  
Daniel Ellsworth  
Jennifer Evans  
Michelle Mills  
Myron Gunsalus  
Shawn Manos  
Liz Schardine  
Heather Smith  
Megan Specht

### **Others Present**

Candice Brannen  
Charlotte Buchanan  
Dennis Dobson  
Randi Gadea  
Daniel Katz

1. Welcome and announcements: Introductions
  - a. Welcome of new members and introduction of people on GoToMeeting.
  - b. Dr Lewis motioned to approve previous minutes (with RVU update on last page), Dr Butler second.
    - i. Previous minutes approved
2. Grants and Special Project updates.
  - a. CDC new contains
  - b. HRSA State Evaluation Grant Award
  - c. CURE SMA grant
  - d. APHL grant- bought freezer
  - e. APHL grant – Status pending.
3. Education update
  - a. Monthly newsletters restarted
  - b. Timeliness tips flyer
  - c. Competencies flyer

- d. Discussion of the best of the best newsletter to call out who is doing well with our state goals.
  - e. Outreach for 3<sup>rd</sup> quarter. 20% of birthing facilities contacted.
    - i. In person contact for 12 facilities (11 on recent western KS)
    - ii. Visit tour 5 facilities
  - f. Tour of Western, KS
    - i. Meet lab/OB/NICU staff at 11 facilities
    - ii. Training Objectives
    - iii. 778 miles in two days. 4 facilities 5 trainings 48 individuals
4. KHEL Update
- a. Continue to be fully staffed.
  - b. Currently analyzing 6 days a week.
  - c. 2019 approximately \$175,000 in grants.
  - d. CLIR data uploaded 2019 (not done since 2014)
  - e. Shobana acknowledged that testing on Saturday is a big deal and a great accomplishment.
  - f. Going Live February 1, 2020 for SMA.
    - i. SMN -2 type send out to Wisconsin State Lab of Hygiene.
    - ii. Heather asked the have a description of SMN and SMA type. An explanation of SMN and type was given.
      - 1. KHEL, Shobana and Dr Zuccarelli clarified that # of SMN-2 copies helps predict severity of SMA
  - g. New SCID cutoffs with SMA Multiplex starting February 1, 2020.
  - h. LSD – Baebies SEEKER installed.
    - i. MPS-1 and Pompe validation started January 2020.
    - ii. Fabry and Gaucher will be used for QC purposes only. Will also help determining if bloodspot specimen has been compromised by heat exposure.
    - iii. Estimated go live in 4<sup>th</sup> quarter of 2020.
    - iv. Baebies suggested a lower cut off for repeat screening as the value decreases as the baby getting older.
    - v. Michelle Leeker stated that every state around KS has legislation to implement Krabbe. Michelle Mills indicated that Krabbe is screened in a different manner and KHEL does not use that testing method.
  - i. Baebies will do second tier testing and will be able to find the following.
    - i. Variant
    - ii. Clinical prediction
    - iii. Variant Effect
    - iv. Severity
    - v. CRIM prediction
    - vi. Zygosity
  - j. In the future Baebies SEEKER will be used for Duchenne Muscular Dystrophy and MPS-II
  - k. Unsats by KHEL. Increase May to June – some of increase due to multiple Unsat codes used by KHEL for specimens.
5. Follow up updates:

- a. Realignment: Education will be for all NBS screening programs (CCHD, Hearing and Bloodspot)
  - b. Data Manager will look thru and go over all NBS data (CCHD, Hearing and Bloodspot)
- 6. KDHE Agency Updates
  - a. Alignment of Blood spot screening and Point of Care.
- 7. Pending abnormal for the year was explained. 41 pending cases for 2019.
- 8. Statewide Timeliness for 2019. Pushing for collection age close to 24 hours.
- 9. Transit time is staying stable but still needs to be improved, currently averaging 72 hours for transit times.
- 10. Collection age is averaging 37 hours, would like to see closer to 24.
- 11. Unsats have trended downward for the year. Both uneven saturation (code 950) and not enough specimen (code 930)
- 12. FP vs. Diagnosed both in the NICU and well-baby.
  - a. False positive breakdown for future meetings.
    - i. **Action item:** When giving FP reports, please break down by Analyte with a note of which disorders the analyte associates with.
- 13. NewSTEPS Quality indicator data report.
  - a. Unsats. Q1 – Unsats (specimen quality)
  - b. Unsat greater than 10 days from collection to receipt by KHEL.
  - c. Q12 Missing essential demographics.
  - d. Follow up timeliness. Only 3 were called out greater than 24 hours.
  - e. Q15b Collection to receipt timeliness is improving slightly.
  - f. NewSTEPS quality indicators
    - i. Q15b receipt to report- means receipt of specimen to final report. improving
    - ii. Q15c Birth to report great than 8 days is decreasing.
    - iii. Q15d birth to report. Staying stable at 75% of the specimens have
    - iv. **Action Item:** Request for Q15c (receipt by KHEL to report date) **broken down for time critical** results will be reported at next meeting.
    - v. Deb asked what region means for lab. Myron stated it means Heartland region.
- 14. CCHD Update.
  - a. F/U has increased the number of screens being reported for 2019.
  - b. 2018 86% screens. 2019 90% screen.
  - c. Reasons not screened. Homebirth, error in charting, discharged, no consent, deceased.
  - d. 2019 CCHD FP and Dx. 98 infants that had failed screen. 35 were data entry error. 5 are pending. 5 other medical condition. 33 were other heart conditions.
  - e. Dr. Katz's asked if screening was improving identifying babies before they become symptomatic. Was the baby symptomatic? Should we be asking this question.
- 15. Lunch Video – <https://www.youtube.com/watch?v=HmYCN0vAepA&t=28s>
- 16. Statute Updates: Heather Smith shared the legislative update. She handed out the legislative bill, KDHE Legislative Agenda-NBS Funding, and KDHE Legislative Agenda- NBS bill.
  - a. History. Why we need this clean up
  - b. What it means for the NBS program going forward. If we are able to expand we will be able to expand our screening.
    - i. Dr. Butler is concerned about removing the word genetics.

- ii. Line 14 on the legislative bill removing the word genetics. Counter discussion was that not all conditions are genetic.
  - iii. Discussion about why KS is not charging for NBS. Concerns as insurance can charge for the screen. Hospital association stated it is not that simple.
  - iv. If the council recommends having a fee for NBS then KDHE will do the work to explore if KS will be able to go to a fee for screen.
    - 1. Michelle Leeker, Dr. Butler, Debra Stern and would like to table that discussion until we know if the legislative bill will be approved to expand the funding.
  - v. **Action Item:** Discussion has been tabled until the May A/C meeting to address if KDHE needs to explore the option for a fee for screen.
  - c. What is needed from the Advisory council? Deborah Stern with the hospital association has asked that the members support the bill as it is presented and not hold out for a fee for screen it will send mixed messages.
  - d. Next Steps. Info briefs, family videos to explain NBS.
17. SMA Overview-
- a. Go live date is February 1<sup>st</sup>, 2020.
  - b. Subcommittee approved all documents and educational materials to be send to the PCP's and families.
    - i. KDHE communications also approved.
18. LSD Overview- Subcommittee is being formed to include specialist, lab staff, and parents.
19. Donna Pitman and husband Cody, have a son Gabriel with Late onset Pompe.
- a. Knowing what their son had and what the expected outcome would be has really helped them prepare for what to expect.
  - b. "Late onset" could mean 7 days 7 years or never.
  - c. Knowing a dx has helped them access a community of support.
20. Comments and Discussions:
- a. SMA notification process. Shobana is concerned that the families want to hear from a specialist that knows the condition. Discussion was that families also like to hear from their own PCP who they are familiar with.
  - b. Discussion about how to educate PCP's on conditions. A/C was advised that the condition specific ACT sheets for PCP and information for families is are available on the KS NBS website.
  - c. **Action Item:** discuss simultaneous notification of PCP and Specialist for SMA at same time.
21. Next meeting is scheduled for May 14<sup>th</sup>. Possible conflicts on the 14<sup>th</sup>, possibly have meeting on 7<sup>th</sup> or 28<sup>th</sup>?