

MINUTES

KS Newborn Screening Advisory Council Meeting
May 16, 2019
Topeka, Kansas

Members Present

Grace Brouillette, MD
Jennifer Gannon, MD
Shobana Kubendran MBBS, MS, CGC
Vance Lassey, MD (phone)
Michelle Leeker
Karey Padding
Deborah Stern
Meghan Strenk
Julie Wellner, RN
Selina Gierer, MD (phone)

Members Absent

Merlin Butler, MD
Kenneth Goertz MD
Michael Lewis, MD
Susan Pence, MD
William Randall Reed, MD
Michelle Ruebke, CPM
Dee Spade, DO
Jean Stork
Jakica Tancabelic, MD

Staff Present

Shawn Manos
Michelle Black
Mary Hinze
Megan Specht
Jan Conklin
Heather Smith
Kayzy Bigler
Jennifer Evans
Myron Gunsalus
Michelle Mills
Emma Bradley
Kathy Modin

Others Present

Dennis Dobson
Jamey Kendall
Bryce Heese, MD
Robert Moser, MD
Rachel Sisson
Andrea Atherton
Patrick Hopkins
Carolina Beltran, MD

1. Welcome and Introduction
 - a. "Why do you come to the A/C meeting?"
 - b. Approval of minutes Shobana motioned and Karri seconded.
 - c. Actioned items from last meeting were reviewed.
2. NBS Program Updates
 - a. General program updates
 - i. Program QI improvements
 - ii. NewSTEPS recommendations
 - iii. Shared Protocols
 - iv. Future Enhancements.
 - b. Current Grants
 - i. NewSTEPS Timeliness Grant
 - ii. CDC New conditions Grant Y2 continuation application
 - iii. HRSA state Evaluation Grant Award
 - c. Education Updates

- i. DOB and TOB Policy Change
 - 1. Info Brief
 - ii. Unsat Relative increase in Standards
 - 1. Bloodspot poster Mailed to hospitals that collected in 2018.
 - 2. Info brief
 - iii. Education Outreach
 - 1. 20 facility site visits approx. 1/3 of the birthing hospitals in KS
 - 2. Monthly Newsletter
 - a. Every Quarter doing a recognition reporting
 - 3. Updated Parent Brochures
 - 4. Moving Forward: Need for increased trust between PCP and Program recommended next steps on referrals
 - d. KHEL
 - i. Fully staffed as of 5-20-2018
 - 1. New staff member 3.5 yr. experience. MS focus.
 - 2. New staff, Phlebotomist Collection focus
 - ii. HPLC new testing.
 - iii. Lab improvements
 - 1. Eval of cutoffs.
 - 2. Developing a more standardized approach to cutoff.
 - 3. Increased in lab turnaround time.
 - e. Follow Up Program Updates
 - i. Losing educator, Personnel is resigning
 - ii. Added Personnel, Megan, to follow up on CCHD and new conditions.
 - iii. Process review – timelines on when to follow up on abnormal notifications.
 - iv. Unsats process trial period.
 - 1. Concerns over what will be missed if you wait 10 days.
 - 2. What is the % of Unsats in the NICU **ACTION ITEM**
 - 3. What is the % of Unsats that are >24 and in the NICU. **ACTION ITEM**
 - 4. Risk management when hospital is not recollecting.
 - 5. Report back to A/C after 30 days of trial. **ACTION ITEM**
 - f. False Positive and Diagnosed Case Report
 - i. 2017 changed from derivatized to non-derivatized method.
 - ii. Made changes to MS cutoffs.
 - iii. KHEL will monitor the FP rate and track results. They will review with Follow up every six months.
 - iv. Patrick reviewed values from KHEL and trends since 2017.
 - v. Results were compared to MO for evaluation.
 - vi. Is it possible to know if the repeat and refer reported are NICU? **ACTION ITEM.**
- 3. NewSTEPS Quality Indicator Data
 - a. Myron presented on Turnaround time for KHEL. TAT.
 - b. Shobana asked if a specimen is received on Friday is it checked in on Friday. Myron confirmed it will be checked in on Friday but there is a cutoff. If the specimen comes in after the time cutoff it will be checked in on Monday. The cut off time is in the morning.

- c. Transport will be the next effort.
 - d. New Logo has been updated.
- 4. Working Lunch
 - a. Pompe News Video was shown to the committee
- 5. Newborn Screening Strategic Assessment
 - a. Data system review – In progress
 - i. Reviewing capacity of existing data system in preparation of expansion
 - b. Fiscal review – in progress
 - i. Reviewing fiscal needs for program as related to expansion and long-term enhancements to the program
 - 1. Complete SMA
 - 2. In progress LSD expansion (eg MPS-1, Pompe)
 - 3. Not Started Leukodystrophy's (XALD, Krabbe)
 - c. Surveys – in progress
 - i. Assessing stakeholder expectations and programmatic education needs.
 - 1. Legislative survey
 - 2. Provider survey
 - 3. Consumer/Parent survey
 - d. PEAS – completed
 - i. Assessing the program to refine and enhance all of the system components.
 - e. Discussion about moving forward and parents advocate voice will be key in funding the program. Doctors acknowledged that they cannot speak to the legislature in regard to funding future expansion.
- 6. PEAS – Document was handed out to address the most significant concerns.
 - a. Education plan
 - i. Making education a priority
 - ii. Consider printing all materials in non-English
 - b. Monitoring of Timely and Universal Screening.
 - i. Create an automated way of comparing birth certificates and blood spots, with a manual audit annually to make sure it is working properly.
 - ii. Follow up will need to develop strong protocols for subsequent screening, obtaining reasons for not completing subsequent screens.
 - c. Personnel
 - i. Develop a plan for recruitment and retention of employees.
 - ii. Assure all training and competency requirements continue to be maintained and reviewed.
 - d. Contingency Plan
 - i. Test the contingency plan on a routine basis.
 - ii. Desktop training for testing can be used.
 - e. Computer Information System
 - i. Need to have seamless system from instruments to follow up
 - ii. System needs to be compatible with outside systems.
- 7. SMA Overview
 - a. Overview- CDC method, analyzed along with SCID

- i. Training and Familiarization. Personnel attended training in Feb 2019
 - ii. Validation and Pre-Pilot Phase
 - 1. Proposed June to July 2019
 - 2. 3000 Blinded Specimens from Missouri
 - 3. Correlation with known positives from CDC and Other States
 - 4. Does not affect Follow-Up or Specialists
 - iii. Full population study
 - 1. Proposed late summer to early Fall.
 - 2. All NBS specimens to KHEL will be tested.
 - 3. Positive screens will be sent to Follow up
 - 4. Very low volume of Presumptive Positives expected
 - 5. 2nd Tier testing for SMN 2 copy number could be considered
 - iv. LIVE Screening and Reporting for SMA
 - 1. System of testing, referral, follow-up and confirmation is working
 - 2. SMA screening results appear on all NBS lab reports
 - b. New Disorders
 - i. LSD screening in Kansas
 - ii. 2 LSD (Pompe and MPS 1) or 4 LSD (Pompe, MPS 1, Gaucher and Fabry) It was stated that if we use the 4 we would be screening outside of the RUSP.
 - iii. Michelle Leeker stated that if you are adding two non RUSP LSDs then there is no reason we can't add Krabbe. Andrea Atherson responded with it is not FDA approved for the babies and is not included in the kit provided.
 - iv. Testing for all 4 (Pompe, MPS 1, Gaucher and Fabry) Increases specificity of the test allowing for more accurate results of Pompe and MPS 1 because Fabry and Gaucher are "well behaved" and we will see less false positives in Pompe and MPS1
 - v. WHY ADDITIONAL LSD's
 - 1. Increases specificity
 - 2. Reduces considerable number of false positives due to
 - a. Heat/humidity
 - b. Diluted/contaminated
 - c. Poor specimen quality
 - d. Low leukocyte counts
 - vi. Showed a slide for FDA approved methods for consideration
 - vii. Is there a PPV for the two different methods? **ACTION ITEM**
 - viii. Wants to know what state labs who are doing MS testing for LSD and what they think. **ACTION ITEM**
 - ix. Every Krabbe case in MO is doing molecular testing.
 - x. African Americans are getting disproportionately associated with high false positive rate. Per Dr. Gannon
 - xi. Further discussed needed in the Metabolic subcommittee
 - c. Subcommittee Michelle gave an update on the SMA subcommittee
8. NewSTEPS Recommended Update -document handled out detailing specific actions.

- a. Heather addressed which activities were completed ongoing, InProgress, future activities, and future discussion.
 - b. Heather address that we will no longer bring completed activities and completed ongoing/ continual activities to A/C but will give progress on in progress/Planned, Future activities, and further discussion Necessary.
 - c. On the document 13-15 Need to be under Further Discussion Necessary and 16-18 need to be under Future Activities (Currently on HOLD)
9. New Business
- a. Disorder champion for program to contact when there is a question.
 - i. Might want to consider a formal consulting agreement.
 - b. More frequent meetings.
 - i. Motion made by Shobana every 4 months and Karey seconded. Approved.
 - ii. Sept 19th is the proposed new date.
 - iii. Have the SMA subcommittee face to face in conjunction with the A/C
 - c. Voting Members
 - i. Letter needs to go to members that have not attended more than 2 consecutive meeting explaining that they may not be eligible to vote or participate due to lack of communication and participation.
10. Adjournment – Shobana needed 2 minutes early with a lot more confidence and trust.
11. No Subcommittees met after the meeting.