

MINUTES

KS Newborn Screening Advisory Council Meeting
May 3, 2018
Topeka, Kansas

Members Present

Selina Gierer MD
Shobana Kubendran MBBS, MS, CGC
Vance Lassey, MD
Julie Wellner, RN
Merlin Butler, MD
Susan Pence MD
Meghan Strenk
Michelle Leeker
Michael Lewis, MD

Members Absent

James Casey, MD
Jennifer Gannon, MD
Jean Stork
Kenneth Goertz MD
Deborah Stern
William Randall Reed, MD
Karey Padding
Jakica Tancabelic, MD

Staff Present

Shawn Manos
Michelle Black
Heather Smith
Caryn Masters
Annie Gile
Elizabeth Schardine
Kayzy Bigler
Jennifer Evans

Others Present

Charlotte Buchanan
Liz Cook
Dennis Dobson
Bryce Heese, MD
Randi Hanson
Kaylee Seyferth
Joy Sherrick
Kate Segal
Carolyn Jones
John Sykora
Liz Fennell

Kansas Newborn screening advisory council May 3, 2018. Held at KaMMCO 10:00am to 2:00pm

1. Welcome and introduction
 - a. On the phone Dr. Shaffer and Dr. Pence.
 - b. Approval of minutes motion to approve Dr. Butler and Dr. Lewis.
 - i. Action items: Could not add NBS to immunization registration.
 - ii. Additional testing options:
 - iii. CF updated: complete: Per Caryn, it is going well, moving on to Phase 2, updating testing of instrumentation. Current method is FDA approved.
 - iv. Hemoglobin: subcommittee. Dr. Tancabelic retired. Need to contact hematologists at Wichita, KU.
 - v. Metabolic subcommittee: secondary reporting.
 - vi. Advisory council satisfaction survey:
 - vii. CCHD: data facilities, KDHE working with Office of Vital Statistics to update requirements.
2. NBS Program Discussion: Program Discussion.
 - a. Presented NewSTEPS report by program intern, Kaylee Seyfrefth.

- i. Michelle Leeker, parent advocate, stated she wanted to see Krabbe added since a lot of other states screen for Krabbe.
 - ii. Shobana, genetic counselor, asked how fees are affected. Heather, KDHE SHS director, replied that funding is looked at each time a condition is considered for the panel.
 - iii. Action Item-Stand point of PPV needs to be added to the table.
 - iv. Action Item-Add to the table for XALD Endo and Neuro along with genetics.
 - v. Action Item-Email this form to the AC so they have it electronically.
 - vi. Action Item-Dr. Butler is asking for a list of instruments from the lab. Can be found on NewSTEPS.
 - vii. NewSTEPS 360 video from Colorado.
 - viii. Changed regs from 24-72 to 24-48. And partnered with NewSTEPS to partner with FedEx to ship specimens.
 - 1. Action Item-Wesley is still getting 20% of their specimen results greater than 7 days from collection. More details on tracking specimens and when it's getting reported.
 - b. KHEL updates. Hemoglobinopathies.
 - i. Plan to go live June 2018
 - ii. UPS Pilot Project
 - 1. Y1 Included 7 birthing facilities
 - 2. Y2 as funding allowing will expand to more facilities
 - iii. Number of specimens that is received greater than 10 days has decreased.
 - iv. Unsat has remained under 3.0%
 - v. Received award for Needle Movers at the NewSTEPS annual conference.
 - vi. Heat map showing that KS does not charge a fee for NBS.
 - 1. Action Item-Dr. Lewis asked for the states that bill for NBS and who they bill? Is it included in the hospital bill?
 - vii. Heat map showing Labs that use Perkin Elmer.
 - viii. Heat map showing number of days lab is open.
 - ix. Heat map showing number of days follow up is open.
 - x. Heat map showing newest conditions that were added to the RUSP by state.
 - xi. Heat map showing number of core disorders screened.
 - xii. NBS how do they categorize disorders?
 - c. Quarterly reports. Wanted the deceased in the abnormal category they are in.
 - i. Can we look at trends in reporting?
 - d. CCHD mandate: Formally added Feb. 2018.
 - e. Training Curriculum: Kaylee showed her training curriculum that is ready for NBS.
- 3. SMA update: Type 1 SMA Can lose 90% of neurons in 6 months leading to significant motor function loss.
 - a. By July 2018, SMA is expected to be signed by Secretary of HHS to FORMALLY be adopted to National RUSP.
 - i. Was formally 'recommended' in February – still needs signed by Secretary.
 - 1. In last 10 years, no test recommended was not signed into RUSP so SMA is coming to National RUSP!
 - b. Spinranza is an antisense oligonucleotide.
 - i. Spinraza makes SMN 2 fully functional.
 - ii. SMA is more common than other disorders on the current RUSP. 6th most common.
 - iii. Action Item-Will have the link to watch videos.
 - iv. Biogen does have a gap filling service to cover Spinraza until insurance covers it.
 - 1. UT, MN, OH, NY currently screening for SMA.
 - v. Want to be able to multiplex with SCID.

- vi. MN did cost analysis – showed that multiplexing SMA with current SCID testing via PCR, only adds at most \$0.15/test
 - vii. Cure SMA, Biogen and CDC helped develop PCR assay. Can detect 99% of 95% of exon 7 deletions.
 - viii. This method CANNOT be multiplexed with PE SCID testing... CAN be combined w/ ANY OTHER PCR testing method for SCID
 - ix. Action Item-CureSMA has a good Algorithm for each type of SMA (0-4) – get copy of this
 - c. John – parent of child w/ SMA advocating for the addition of SMA
 - i. Son is on Spinraza but was not originally when diagnosed. Spinraza not available at time of his diagnosis.
 - 1. Son is making improvements but may not establish 100% mobility
 - d. Dr. Lassey asked if Biogen can purchase equipment for lab
 - i. Biogen has encouraged CDC to allot \$ to help states implement new conditions, not only SMA
 - e. Dr. Brad Shaffer, Professor of Genetics and Pediatrics at the University of Arkansas for Medical Sciences contracts with tele-genetics clinic in Wichita discussed State Genetics Plan.
 - i. Biggest impact that Dr. Shaffer has seen was adding the availability of the lab. Increasing the testing days. Decreasing the turnaround time.
 - ii. Adding telemedicine in Wichita and Overland Park.
 - iii. Will work as adjunct professor at KUMC.
 - iv. Goal to finish Project by 2019. Adding Genetic services in KS.
4. Sub-Committee Reports
- a. SCID – not very many cases sent to Quest (~12), no True cases found yet. Selina would like to rework a way she is notified b/c currently they are contacting her via phone. Issue when she's away from office.
 - i. Action Item-Need to re-work algorithms to include very small BW babies (<500g) – Selina will work on this
 - ii. Action Item-Need to re-work algorithms to include babies w/ specimens that have at least 1 normal screen – KS will start NOT calling SCID abnormal if baby has ANY normal SCID screen
 - iii. KS has high consanguinity with SCID in a particular region in KS. In 4 years, Selina confirms there have been 5 confirmed cases of SCID in Kansas.
 - b. 17-OHP False Positive
 - i. New progesterone treatments for mothers with pre-term births are potentially leading to an increase of false positives.
 - ii. Possible research study by KDHE to investigate the FP, injection link.
 - 1. May investigate vital records to see if charted as mother receiving the 17-P treatment if info is available.
 - 2. Dr. Butler asked about the half-life of the injections, 7 days vs 2 weeks could make a difference.
 - iii. If link found, could result in updating/changing the algorithms we use for 17-OHP similar to what we do for TPN.
5. New Business
- a. Elections moved to the end of meeting per Dr. Lassey
 - b. Strategic Planning – Heather Smith, KDHE SHS director, spoke about strategic planning with the idea of where we want to go in the future
 - i. Doing an overall Strategic Plan & evaluation of the NBS Program as a whole.
 - 1. Do a Complete Overview to prioritize the importance of adding new conditions, role of lab, F/U, capacity to do testing, the laws themselves.

2. Proposal to agency to add another FTE in F/U to do some of this work and get ahead of the curve before any conditions are added or program expands.
 3. Shobana asked if we can look at updating the laws to allow NBS to charge like other states.
 - a. Heather, KDHE director, says so far, funding from Fee-Fund has been sufficient but charging can be included. Won't be a focus but can be considered as part of the plan.
 4. Dennis Dobson, community member, mentioned that if AC makes formal recommendation to look into expanding, looking at the laws and doing the Strategic Planning then KDHE can formally look into it
 5. Wants to look at what's BEST for the NBS Program and not be limited
 - a. Dr. Shaffer would like to help with linking into the State Genetics Plan.
- ii. Expanding the Panel
1. Dr. Lassey brought up the topic to Council for a formal recommendation to add
 - a. Asked CMH about their expertise b/c of work with MO who is already screening
 - i. Dr. Hesse mentioned POMPE would detect late onset cases as well.
 - ii. He is NOT against screening for POMPE.
 - iii. MPS-1 & POMPE both get a lot of False Positives but despite that, he recommends it's an overall good thing.
 - iv. Says X-ALD is also a good disorder to add but MO does not screen for that yet.
 - v. Dr. Hesse mentioned KHEL will need expertise and info so that they use the best equipment to help reduce the FP's. Can ask CMH for help.
 - vi. While KHEL has the MS/MS instrumentation to add disorders to panel, kits, reagents and staffing are NOT available yet.
 - b. Michelle Leeker asked for addition of Krabbe in addition to other 4 on NewSteps review table presented by program intern.
 - i. Dr. Hesse withheld comments about Krabbe because of how rare the disorder is (1/500,000) incident rate and the number of FP's of this as well.
 1. Detected 1-2 since 2013 in MO population
 - ii. Also mentioned the effectiveness of how well the bone marrow treatments work and wasn't sure how well those worked.
 1. Leeker understood Dr. Hesse's hesitation.
 - iii. Dr. Lewis mentioned that this is one of the advantages of doing the robust Strategic Planning. That the plan will help establish the "just because we can, should we" argument!
 - iv. Dr Lassey said it's very valuable to have MO and their experience to ask questions like this.
 - v. Heather asked about Krabbe being added to the uniform panel. It has not been tried to be added since 2009. Each state has to add it on its own panel.
 - vi. Action Item-Dr. Heese asked that someone from KDHE would go to Hunter's Hope this summer in hopes of getting more information on Kabbe disease. July 18 in NY.

- vii. KDHE can pay for genetic services if family applies and qualifies for funding.
 - viii. Using genetic counselors and knowing what we can expect from them and what to expect from NBS.
 - c. Dr. Mike Lewis made a motion to add all 4 conditions to the panel and Shobona 2nd it.
 - i. Strategic planning should include Krabbe.
- 2. Elections:
 - a. Dr. Lassey nominated Shobona for chair and Dr. Geier 2nd. Shobona accepted and Jen was nominated for vice Chair.
 - b. Action Item- Jen will be asked if she accepts over email.
- 3. Motion to adjourn was made and seconded.