MINUTES
KS Newborn Screening Advisory Council Meeting
October 26th, 2017
Topeka, Kansas

Members Present
Selina Gierer MD
Shobana Kubendran MBBS, MS, CGC
Vance Lassey, MD
Julie Wellner, RN
Merlin Butler, MD
Susan Pence MD
Jean Stork
Jennifer Gannon, MD
Meghan Strenk

Members Absent
James Casey, MD
Kenneth Goertz MD
Deborah Stern
William Randall Reed, MD
Michelle Leeker
Karey Padding
Jakica Tancabelic, MD

Staff Present
Shawn Manos
Michelle Black
Heather Smith
N Myron Gunsalus
Caryn Masters
Kelley Hale

Others Present
Annie Gile
Elizabeth Abbey
Dennis Dobson
Laurie Gwyn, MD
Geri McFall
Bryce Heese, MD
Randi Hanson
Shalae Harris
Ingrid Larson
Carolina Beltran, MD

- Approval of previous meeting minutes
    - Jen Gannon 1st and Mike Lewis 2nd
- New steps site visit report recommendations. Go live date is Nov. 6th. We will be picking up a consultation to help with medical and lab perspective. We are meeting most of the targets. Lab will be looking at the indicators. Will look at turnaround time. Asking for feedback for recommendations as well.
- LIMS update: now have the ability to give secure email reports. Office must provide a secure email account. Testing new LIMS system that could be available beginning of 2018. This system will be cloud based. Question was asked if they can get it emailed and faxed and the answer was no they have to choose one. The emails do come as a batch. They get reported when the batch is ready. It could be 4-6 times a day when the batch is ready. There is a fillable PDF on the KDHE NBS website. Dr. Lassey recommended a secure data base where if they have a secure data base they can access the information. MO has a system where providers can be given access to log in and review reports.

- Action Item - Find out who has access? Do physicians or just specialist?

- Timeliness: Project to improve timeliness. Working on ways to have a point of contact, and not batch specimens. Having visualization to help collect. Related to mailing of specimens. Lab is tracking time of receipt to time of reporting. They are averaging two days. Dr. Bloom's concern is reporting. Wesley will directly compare data with the lab to find the discriptiony. 7 facilities have a contact with UPS to overnight the specimen.
- Lab update: lab is fully staffed.
    - Values have now been provided to specimen gate software.
    - Change from high, moderate or low to repeat or refer.
    - Lue level is being increased.
    - HCY will look at birth weight.
    - VLCAD level is going up.
    - CUD is going down and a secondary analyte is being added.
    - GA-1 raised level
    - C3 will have two cut offs look at the babies age in hours. One cut off less 144 hours.
    - SCID update Birth weight dependent
    - UPS Pilot Project April 2017 to April 2018
    - NBS Forms updated.
    - Perkin Elmer is going to train staff on instruments on site in November.
    - PE is going to meet w/ KHEL and APHL about service issues from PE side. Shobana and Selena will be willing to attend to voice concerns.
    - Perkin Elmer is providing training for one staff member to the Indiana state lab in February regarding specimen gate software.

    - Future projects: Variant Biorad for HBG results are less subjective. Less time intensive for analyst. Will quantify the HBG percentage and allow for sending of chromatograms.
        - Shobana offered assistance if needed from hematologists
    - GSP for Galt, Bio, TSH, IRT, OHP
        - Will be reagent rental contract but upfront cost is expensive
- FU updates
        - Michelle full time, educator still vacant
        - Heather discussed briefly about the strategic planning for the program and that we'll be seeking advice from the AC
        - New Brochures are available on the website.
- CF updates
        - Dr Lewis discussed patient that invoked change in algorithm
        - Discussed his new proposed algorithm and why it would save a lot of time for FU on CF kiddos
        - Need to change the wording in the letter and sit down with each family individually to explain R117. 5T7T

- Action item: Update the algorithm and add the two CF centers to the website. Add link to accredited CF center. Reevaluate the report.

    - State genetics plan: Dr. Shaffer looking at stake holder meeting. KU and KDHE will look state plan.
    - Telegenetics grant from Heartland. First sight is Hays. Shobana will be the genetic counselor and Dr. Beltran will be available.

- Action Items: hemoglobin subcommittee will be developed and let KDHE know if you would like to be a part of it.

- Action Items: Kdhe will draft a survey to see if A/C members are getting what they need meeting 2 times per year.
    - Heather will send out 4 questions that will guide where the A/C will go next.
    - Give them back information on what is on the horizon.

- SCID update: is there going to be a news release? Have a family that would be willing to speak on behalf of announcements of SCID release.

- Action item: CCHD was asked to send data out data for facilities that are screening and reporting.

- Spinal Muscular Atropy- SMN1 caused by a mutation in survival motor neuron gene 1 (SMN1) gene. Without the SMN protein, the nerve cell dies.
    - Genetic recessive- both are carriers.
    - No muscles but they feel pain.
    - Affects muscles NOT minds
    - Hearing and sensory intact-feel pain
    - Average to above average IQ
    - Does NOT distinguish between race or gender
  - Incidence 1 in 10,000 to 1 in 11,000 births
    - 4 births a year born in Kansas
  - Number one genetic killer of children under 2 years of age
  - 1 in 40-60 people are carriers
  - Types of SMA
    - Type 0 usually die in utero or several weeks after birth from respiratory failure.
    - Mother may notice limited fetal movement
  - SMA Type 1 most common
    - Rapid onset
    - Floppy baby
    - Never walk
    - Lose swallow reflex
    - Bellshaped chest
    - High incidence of respiratory arrest
    - Most die before their 2nd birthday
  - SMA type 2
    - DX age 6- 24 months
    - Can sit independently
    - Never walk
    - Couunicate freely
    - May need respiratory support
    - Tolerate regular diet
    - Confined to a wheel chair
    - Scoliosis usually by 6 years age
    - Life expectancy into 30's
  - SMA type 3
    - Usually dx between 18 months to 3 years sometimes not diagnosed until teenage years.
    - Lose ability to walk as grow
    - Clumsy child
    - Regular diet
    - Normal life expectancy
  - SMA type 4
    - Adult onset – 35 years

        - Muscle weakness worsens
        - Normal life span
    - Early diagnosis
    - NBS is key to successful intervention and treatment.
    - Respiratory support
    - Ot/pt
    - Diet n/g tube/g-tube/ weight maintence
    - Medication to slow down progression/ halt disease progress.
    - Spinraza (Nusinersen)
        - Simulates the SMN2 back up gene
        - Administrered intrathercal injection lumbar puncture
        - Dose  12 mg per admistration by a health care provider
        - 4 loading doses.
        - Maintenance does is given once every 4 month after that.
        - Cost. $125,000 per dose. ($500,000 - $625,000 first year and $325,000 each year after.
    - Funding for Spinraza
        - Primary insurance company first
        - Biogen SMA360 (benefit)
        - KU is the only hospital in Kansas that is approved to adminitster Spinrzaz in the State of Kanas. (Children's mercy Hospital in Kansas City, Mo accepts Kansas Medicaid)
    - Community support
        - Cure SMA (Website) Has a Kansas city Chapter includes KS, MO, OK NB
        - Social media
        - Jadonshope.org based in Olathe, KS
- MO is the first state to test for SMA. It is multi plexed with SCID PCR looking for SMA1 and SMA 2 we would have to validate SCID again. SMA is not on the RSUP as of this meeting.
    - To add a recommendation to the panel a committee is made and researches the cost to test, equipment, and long term follow up while doing a cost analyses. We are not required to screen what is on the RUSP. How to you fund it and how to you fund it if you find something?

- Action Item: What tests can be run on the instruments that we are already using with the amount of staff that we have?

- Quarterly reports
    - CAH reports missing
    - Blood saturation- could have more training and practice
        - 3 year low for unsats but our numbers are going up
- New business

    - RUSP list that is current.
- Elections at next meeting